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MARCELA BEZERRA DIAS

**O ACONSELHAMENTO DOS FAMILIARES APÓS O DIAGNÓSTICO DE
CARDIOPATIA CONGÊNITA DO FETO: REVISÃO DE ESCOPO**

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Dissertação apresentada ao programa de Pós-Graduação em Ciências da Saúde da Universidade Federal do Amazonas, como requisito para a obtenção do título de Mestre em Ciências da Saúde, na linha de pesquisa em pesquisa clínica e saúde pública.

ORIENTADORA: PROF.^a DR.^a LUCIANE ALVES DA ROCHA AMORIM

MANAUS

2023

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Aprovada em: 12 de Abril de 2023.

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Aos profissionais que publicaram as suas pesquisas e contribuíram para novas descobertas, àqueles que desejam humanizar o seu trabalho e, acima de tudo, aos familiares com diagnóstico de cardiopatia fetal que necessitam de um acolhimento e aconselhamento para o tratamento de seus bebês.

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Respeitosamente, a todos vocês.

Quando percebem que foram profundamente ouvidas, as pessoas quase sempre ficam com os olhos marejados. Acho que na verdade trata-se de chorar de alegria. É como se estivessem dizendo: "Graças a Deus, alguém me ouviu. Há alguém que sabe o que significa estar na minha própria pele. Carl Rogers (1980, pg.7).

RESUMO

Introdução: Estima-se que 1 em cada 100 bebês nascidos vivos tenha algum tipo de cardiopatia congênita. No Brasil, em torno de 28,9 mil crianças nascem por ano com cardiopatia congênita (1% do total). Dessas, 80% necessitarão de algum tipo de intervenção cirúrgica. O diagnóstico precoce, o aconselhamento familiar adequado e um bom planejamento do parto são fatores que favorecem um melhor resultado terapêutico. Como os dados relativos à prática do aconselhamento desses familiares desde o diagnóstico até o nascimento encontram-se esparsos na literatura, nosso objetivo foi avaliar e entender a extensão do conhecimento científico sobre o aconselhamento familiar no momento do diagnóstico intraútero, assim como identificar lacunas neste campo. **Método:** Esta é uma revisão de escopo que incluiu estudos primários, quantitativos e qualitativos, e que incluíram características dos participantes, amostra, resultados do estudo e os detalhes do aconselhamento como identificação do profissional que realizou o aconselhamento aos familiares e o tipo de aconselhamento familiar realizado. A pesquisa foi conduzida de acordo com as diretrizes encontradas no manual de revisores do Joanna Briggs Institute de Estudos Observacionais em Epidemiologia. A estratégia de busca foi identificada por uma pesquisa sistemática realizada durante o mês de Setembro e Outubro de 2022. Foram utilizados 6 bancos de dados: Medline, Embase, Lilacs, Scielo, Scopus e Web of Science, além da literatura cinzenta: Psycinfo e google acadêmico. O projeto foi cadastrado no banco de dados de registros no Open Science Framework- OSF .O processo de identificação dos estudos foi realizado por dois revisores, independentes, e segmentado em fases: identificação, triagem e inclusão. A análise da qualidade metodológica e o risco de viés foram realizados com a ferramenta de avaliação crítica do Joanna Briggs Institute- JBI. A triagem foi independente de todos os artigos, inicialmente apenas com base no título e no resumo e, posteriormente, com base no texto completo utilizando o sistema Rayyan para a coleta. **Resultados:** Do total de 3719 estudos analisados, vinte e um foram incluídos para a análise. **Conclusão:** Evidenciamos que os estudos de aconselhamento foram realizados exclusivamente em países desenvolvidos, o que gera questionamentos sobre esta ferramenta em uma população com recursos escassos. Ainda, podem ser realizados estudos validando os questionários utilizados para se identificar o grau de compreensão dos pais sobre a doença. Assim como se identificar se existe diferença deste grau de compreensão se a informação for passada por um cardiologista pediátrico ou um médico fetal ou ultrassonografista. **Registro de protocolo:** 10.17605/OSF.IO/7WK45.

Palavras-chave: Aconselhamento, cardiopatia congênita, coração do feto.

ABSTRACT

Introduction: It is estimated that 1 in every 100 babies born alive has some type of congenital heart disease. In Brazil, around 28,900 children are born each year with congenital heart disease (1% of the total). Of these, 80% will need some type of surgical intervention. Early diagnosis, adequate family counseling and good birth planning are factors that favor a better therapeutic outcome. As data on the practice of counseling these family members from diagnosis to birth are sparse in the literature, our objective was to assess and understand the extent of scientific knowledge on family counseling at the time of intrauterine diagnosis, as well as to identify gaps in this field. **Methods:** This is a scoping review that included primary, quantitative and qualitative studies, and which included characteristics of the participants, sample, study results and the details of counseling such as identification of the professional who provided counseling to family members and the type of family counseling accomplished. The research was conducted according to guidelines found in the Joanna Briggs Institute of Observational Studies in Epidemiology reviewers manual. The search strategy was identified by a systematic search carried out during September and October 2022. Six databases were used: Medline, Embase, Lilacs, Scielo, Scopus and Web of Science, in addition to the gray literature: Psycinfo and google academic. The project was registered in the records database in the Open Science Framework-OSF. The process of identifying the studies was carried out by two independent reviewers and was divided into phases: identification, screening and inclusion. Analysis of methodological quality and risk of bias were performed using the Joanna Briggs Institute-JBI critical assessment tool. The screening was independent of all articles, initially only based on the title and abstract and, later, based on the full text using the Rayyan system for collection. **Results:** Of the total of 3719 studies analyzed, twenty-one were included for the analysis. **Results::** Of the total of 3719 studies analyzed, twenty-one were included for the analysis. **Conclusion:** We showed that counseling studies were carried out exclusively in developed countries, which raises questions about this tool in a population with scarce resources. Still, studies can be carried out validating the questionnaires used to identify the degree of parents' understanding of the disease. As well as identifying whether there is a difference in this degree of understanding if the information is passed on by a pediatric cardiologist or a fetal doctor or sonographer. **Protocol registration:** 10.17605/OSF.IO/7WK45.

Keywords: Counseling, congenital heart disease, fetal heart.

APRESENTAÇÃO

A minha trajetória pessoal e profissional vai ao encontro desta rica oportunidade de aperfeiçoar-me dentro do mestrado e especificamente dentro desta pesquisa sobre aconselhamento. Efetiva na área de educação, também autora e escritora de livros de autoajuda, palestras e projetos sociais, psicóloga formada há 15 anos na UNISAL– Universidade Salesiana de Lorena/SP; e estudante da Bíblia Sagrada, vivencio muitos contextos onde o aconselhamento é fundamental para ajudar a consciência situacional, tomadas de decisões, acolhimento e no processo de psicoeducação do ser humano.

Especialista em psicopedagogia, psicologia positiva e coaching, e MBA em gestão de comportamento organizacional pude percorrer experiências profissionais, como, oficial do exército técnico, temporária na área de psicologia, psicopedagogia do Colégio Militar de Manaus e aviação do exército. Peacekeepers integrante da MINUSTHA (Missão das Nações Unidas para Estabilização no Haiti) por ter operado como psicóloga no BRABAT-26, (26º Contingente do Batalhão Brasileiro de Força de Paz), colaborando com a nobre e intensa missão de paz da ONU, de proporcionar um ambiente seguro e estável ao Haiti.

No meu convívio junto à equipe de saúde, pude conhecer uma excepcional médica que desempenhava o seu papel com elevado comprometimento e dedicação muito além do que se era mandatário. Pude perceber que ela estava com sintomas associados à síndrome de Burnout e busquei aproximar-me com o intuito de oferecer uma acolhida. Ela era muito introspectiva para o convívio mais próximo, porém, por estarmos em confinamento e contato diário 24 horas, em alguns momentos este contato era possível até que um dia relatou algo de sua experiência profissional que havia marcado profundamente um parto em um local inóspito de uma mãe que vivia em condições de vulnerabilidade social, acarretando infelizmente o óbito do bebê poucos dias depois do seu nascimento. Este acolhimento também me marcou enquanto pessoa e profissional. Surgiu uma inquietação do quanto temos feito para ajudar na qualidade de vida dos profissionais de saúde? Percebi o quanto a psicologia ainda precisa se integrar para se tornar uma ajuda efetiva para estes profissionais. Também, em algumas oportunidades durante visitas aos pacientes nos hospitais, pude perceber que não existia um protocolo bem

estruturado na condução do aconselhamento e acompanhamento psicológico, para que subsidie a equipe de saúde na comunicação e intervenção com os mesmos. Dados que pude retomar no desenvolvimento desta pesquisa em contato com a minha orientadora que é professora e médica cardiologista e tem me ensinado como ser efetiva na contribuição científica.

Na oportunidade que tive como docente e coordenadora do curso de psicologia na ULBRA/Manaus- Universidade Luterana do Brasil campus de Manaus, busquei compartilhar com os acadêmicos esses saberes a fim de torná-los multiplicadores de ações mais assertivas. A necessidade da atualização científica no campo da psicologia da saúde deve ser o esforço coletivo na construção e compartilhamento dos saberes, tendo como foco a melhoria da qualidade de vida dos pacientes e dos profissionais. Agentes de cuidado, transformação e informação. O caminho é longo, porém deve ser percorrido com muita serenidade e fé, considerando todos os estudos construídos até aqui. Portanto, o tema deste trabalho, aconselhando aos familiares após o diagnóstico de cardiopatia do feto, foi escolhido por ser um problema de saúde muito frequente entre a população gestante e campo onde os profissionais de saúde desempenham o papel imprescindível para orientar as ações do diagnóstico ao tratamento.

O trabalho será apresentado em forma de dissertação e, posteriormente, em formato de artigo científico.

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LISTA DE ABREVIATURAS E SIGLAS

CC	Cardiopatia Congênita
DCC	Doença Cardíaca Congênita
LILACS	Literatura Latino-americana e do Caribe em Ciências da Saúde
JBI	Joanna Briggs Institute
MEDLINE	Medical Literature Analysis and Retrieval System Online
MeSH	Medical Subject Heading
OMS	Organização Mundial da Saúde
OSF	Open Science Framework
PCC	Problema, Conceito, Contexto
PPGCIS/UFAM	Programa de Pós-graduação em Ciências da Saúde/ Universidade Federal do Amazonas
PRISMA-ScR	Preferred Reporting Items for Systematic Reviews and Meta-Analyses extension for Scoping Reviews
SCIELO	Scientific Electronic Library Online
UFAM	Universidade Federal do Amazonas

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1 INTRODUÇÃO

A doença cardíaca congênita (DCC) é a causa mais comum de malformação congênita no feto, afeta aproximadamente 1 em cada 100 recém-nascidos (ROCHA *et al.*, 2013; KOVACEVIC *et al.*, 2022), sendo a principal causa de morte por malformações no primeiro ano de vida (CHAOUI *et al.*, 2012). Segundo a Organização Mundial da Saúde (OMS), a incidência de cardiopatias congênitas varia entre 0,8% nos países desenvolvidos e 1,2% nos países em desenvolvimento, sendo 1% de incidência o habitualmente aceito para o Brasil e demais países da América Latina. Dessa forma, visto que o Brasil registra anualmente 2,8 milhões de nascidos vivos, pode-se estimar o surgimento de quase 29 mil novos casos de cardiopatias congênitas ao ano (CANELO *et al.*, 2012). Destes, cerca de 30% evoluem para óbito ao nascimento e 6% não sobrevivem até o primeiro ano de vida (SIMIONI *et al.*, 2012).

O diagnóstico precoce da DCC pela ecocardiografia fetal mudou drasticamente a medicina perinatal, permitindo um melhor monitoramento fetal, encaminhamento, planejamento do parto, opção de intervenção fetal e manejo pós-natal apropriado (SEWELL; KEENE, 2018). Entretanto, há de se ressaltar o impacto psicológico que este diagnóstico pode ter na mãe e na família (SKLANSKY, 2002). Os estudos são unânimes em reconhecer o impacto emocional desse momento em situações de anormalidade fetal (GOMES *et al.*, 2010). Ainda, neste momento do diagnóstico, os pais podem necessitar tomar decisões difíceis como por exemplo, interrupção ou não da gestação (GENDLER *et al.*, 2021). Neste contexto, a orientação adequada aos pais sobre o tipo de DCC é fundamental. Esta abordagem é conhecida como aconselhamento em saúde.

O aconselhamento em saúde é uma intervenção que consiste em ajudar o paciente a manter ou melhorar a sua saúde em instâncias que vão desde a prevenção à adaptação da doença.) Santos *et al.* (2014) apresentaram uma revisão sobre as abordagens do aconselhamento e da psicoterapia, onde apresentaram o aconselhamento psicológico como dinâmico e importante para a expressão e elaboração de uma comunicação compreensiva. Rogers *et al.* abordam o papel do psicoterapeuta na sua relação com o paciente, baseando-se no fato do relacionamento construído ser esclarecedor (ROGERS *et al.*, 2001).

1.1 JUSTIFICATIVA

No diagnóstico fetal de DCC, o aconselhamento ajuda o familiar a entender o problema, minimizando o impacto psíquico da família na evolução da gestação e contribuindo para a melhor estratégia de tratamento. Os pais podem estar em posição de tomar uma decisão crucial sobre o manejo da gravidez com base nas informações recebidas, por isso é vital que o conselheiro seja realmente capaz de se comunicar com eles, qualquer que seja seu nível de compreensão (ALLAN; HUGGON, 2004; KOVACEVIC *et al.*, 2018). Aconselhar os pais, após o diagnóstico da DCC, é uma tarefa tão importante para o cardiologista fetal, quanto a habilidade envolvida em obter um diagnóstico preciso (ALLAN; HUGGON, 2004). Os efeitos da prática terapêutica do aconselhamento podem ser positivos, pois podem impactar na aceitação da anomalia e na motivação dos familiares para o tratamento da cardiopatia e promover o esclarecimento de dúvidas e de diversos questionamentos. Assim sendo, o aconselhamento familiar eficaz deve fazer parte de qualquer programa de cardiologia fetal. No entanto, são escassos os estudos sobre as técnicas mais eficazes, assim como os dados sobre a avaliação empírica do aconselhamento e sua eficácia (KOVACEVIC *et al.*, 2021).

Nesse contexto, essa revisão de escopo irá examinar e mapear as publicações sobre a prática do aconselhamento dos familiares que recebem o diagnóstico de doença cardíaca congênita no período gestacional, utilizando a metodologia de Joanna Briggs Institute (JBI), conforme descrito no manual de revisores do JBI de 2014.

1.2 OBJETIVOS

1.2.1 Objetivo geral

Identificar as lacunas existentes na literatura sobre o aconselhamento familiar nos casos de diagnóstico de cardiopatia congênita ainda no período gestacional.

1.2.2 Objetivos específicos

Caracterizar os desenhos de estudo encontrados sobre o aconselhamento familiar nos casos de diagnóstico de cardiopatia congênita ainda no período gestacional.

Identificar evidências de que o aconselhamento familiar auxilia no processo de compreensão da cardiopatia congênita do feto.

Identificar se existe influência no processo de compreensão da cardiopatia congênita do feto em relação ao tipo de profissional de saúde que realizou o aconselhamento familiar.

Identificar algum instrumento para a realização do aconselhamento familiar nos casos de cardiopatia congênita no feto.

2 REFERENCIAL TEÓRICO

2.1 CONCEITO DO ACONSELHAMENTO FAMILIAR

O aconselhamento familiar no pré-natal para DCC tornou-se essencial, à medida em que vem aumentando a frequência de detecção (GEDİKBAŞI *et al.*, 2011). É um processo no qual um técnico utiliza competências específicas para ajudar o utente a lidar mais eficazmente com a sua vida (TRINDADE; TEIXEIRA, 2000). É uma das principais tarefas para cardiologistas fetais ou especialistas em medicina materno-fetal (MFM) após o diagnóstico de DCC no feto (KOVACEVIC *et al.*, 2021).

Trata-se, portanto, de uma habilidade complexa que requer conhecimento profundo da fisiologia cardíaca normal e das DCC, dados de tratamentos e prognósticos, capacidade de comunicação e inteligência emocional. Permeia em descrever a doença cardíaca, o curso esperado do bebê, os resultados típicos, as associações genéticas e transmitir incertezas em relação aos resultados do pré e do pós-natal. (KEELAN *et al.*, 2022). Afirmando assim, a profundidade que envolve o saber profissional e o aconselhamento aos pacientes.

Estudos que discorrem sobre a regulamentação abortiva apresentam que por exemplo, na Suécia, sendo o aborto induzido, fica disponível mediante solicitação até 18 semanas completas de gestação e em gestações posteriores após a aprovação do Conselho Nacional de Saúde e Bem-Estar (CARLSSON *et al.*, 2020).

Atualmente, a interrupção por uma anormalidade cardíaca congênita grave e/ou sofrimento materno significativo pode prosseguir para vinte e duas semanas, mas foi mais recentemente estendida para vinte e quatro semanas com a necessidade de um comitê hospitalar especialmente convocado para considerar e decidir sobre qualquer solicitação posterior a vinte, ou mesmo vinte e duas semanas de gestação (MENAHEM, 2012). Normalmente, abortos induzidos após 12 semanas de gestação completas são realizados como abortos medicamentosos e expulsão vaginal do feto (CARLSSON *et al.*, 2020).

Em seu estudo, Lee (2017) apresenta uma tabela contendo elementos importantes para abordar o aconselhamento pré-natal, e a sugestão de como utilizá-los, por exemplo, elementos que versam sobre as informações detalhadas do

diagnóstico considerando possíveis complicações do caso e quais ações serão necessárias. Estes modelos subsidiam acompanhamentos mais assertivos por retratar aspectos importantes que devam fazer parte do tratamento do paciente.

Abaixo será apresentada uma proposta de modelo para a abordagem com a paciente e familiar (Quadro 1).

Quadro 1 – Checklist para o Aconselhamento familiar no pré-natal em casos DCC fetal

Aconselhamento pré-natal biopsicossocial na DCC
<p>Aspectos Biológicos:</p> <ol style="list-style-type: none">1) Esclarecimento médico sobre o diagnóstico.2) Esclarecimento médico sobre o acompanhamento.3) Esclarecimento médico sobre o tratamento.4) Preparo para os desfechos favoráveis e desfavoráveis no caso de evolução ou interrupção da gestação.5) Sugestão de tratamento preventivo para novas gestações.
<p>Aspectos Psicológicos:</p> <ol style="list-style-type: none">1) Anamnese e acolhimento pós-diagnóstico.2) Verificação do grau de compreensão e adesão às informações médicas sugeridas no caso.3) Verificar a estrutura psíquica para o tratamento e para a vivência dos desfechos do caso.4) Suporte emocional de acompanhamento para o paciente e familiar.5) Aconselhamento para a equipe multidisciplinar sobre o acompanhamento do caso.
<p>Aspectos Sociais:</p> <ol style="list-style-type: none">1) Fornecer materiais escritos sobre diagnóstico e tratamento DCC.2) Fornecer informações sobre palestras e grupos de apoio.3) Verificar as questões familiares: financeiras e apoio psicológico.4) Para a alta hospitalar verifica os medicamentos, a necessidade de monitoramento domiciliar e acompanhamento cardiológico.5) Nas questões de longo prazo considerar intervenções cirúrgicas e o acompanhamento cardiológico nas demais fases da vida.

Fonte: Elaborado pela autora.

Estes elementos supracitados ajudam os médicos e os profissionais da equipe multidisciplinar, como os psicólogos e assistentes sociais, na condução do aconselhamento dos familiares, percorrendo diversas dimensões de manejo desde a orientação técnica da doença, até requisitos básicos e complexos que se esperam promover no enfrentamento à situação de crise, o essencial acolhimento emocional.

O aconselhamento aos familiares com DCC pré-natal e seus benefícios é considerado um passo fundamental (RAKHA, 2020). A condução do processo prognóstico, apresentado pelo profissional de saúde, requer ferramentas apropriadas que possam apoiar os pais na compreensão da natureza desta anormalidade e suas prováveis consequências imediatas e tardias. Sem dúvida,

esse aconselhamento precisa ser feito de forma a levar em conta o frágil estado emocional em que os pais se encontram (MENAHEM, 2012). De fato, não é um problema isolado quando citamos as relações parentais, pois se a mãe toma essa decisão sozinha, seja sem o seu parceiro, em um relacionamento oficial ou não, seja considerando a extensão familiar, como avós, tias, tios e irmãos, a verbalização é fundamental e deve ser considerada para que a decisão seja assertiva, precisa e clara. As suas opiniões e posicionamentos devem ser levados em consideração, principalmente se um ou mais membros próximos aos pais afetados têm uma opinião muito forte sobre como a gravidez deve prosseguir? (MENAHEM, 2012).

No caso específico do aconselhamento psicológico, trata-se de uma intervenção do especialista da saúde, que se diferencia da intervenção clínica em aspectos específicos, tais como: ter caráter situacional; ser centrado na resolução de problemas do sujeito; ser uma intervenção focada no presente; ter uma duração mais curta; ser orientada para a ação mais do que para a reflexão (FERNANDES, 2015). Consiste numa relação de ajuda que visa facilitar uma adaptação do sujeito à situação em que se encontra, através da otimização dos seus recursos pessoais, do seu autoconhecimento, das suas capacidades de autoajuda e de autonomia no confronto com as suas dificuldades e problemas (TRINDADE; TEIXEIRA, 2000).

Existem diversas perspectivas teóricas do aconselhamento psicológico, como: psicodinâmicas, humanistas, cognitivo-comportamentais, feministas, construtivistas, fenomenológico-existenciais e sistêmicas. A perspectiva cognitivo-comportamental é a mais recomendada aos contextos de saúde e doença, porque se adapta melhor ao contexto e ritmo próprio da prestação dos cuidados de saúde (FERNANDES, 2015).

A atividade cognitiva do paciente após receber o diagnóstico pode influenciar seu comportamento e suas emoções, podendo acarretar em transtornos psicológicos que são decorrentes das distorções cognitivas – maneira pela qual o paciente compreende os acontecimentos e faz dele suas crenças centrais pouco adaptativas e invariáveis às mudanças. Para tanto, os autores assinalam que a teoria cognitivo-comportamental pode auxiliar o paciente a aderir ao tratamento, oferecendo apoio emocional e qualidade de vida (SOUSA *et al.*, 2015). Desta forma, o objetivo é mostrar que a influência sobre nós não são diretamente os acontecimentos e situações diárias, mas sim a forma que interpretamos cada uma dessas situações, para que possamos agir de forma consciente elaborando

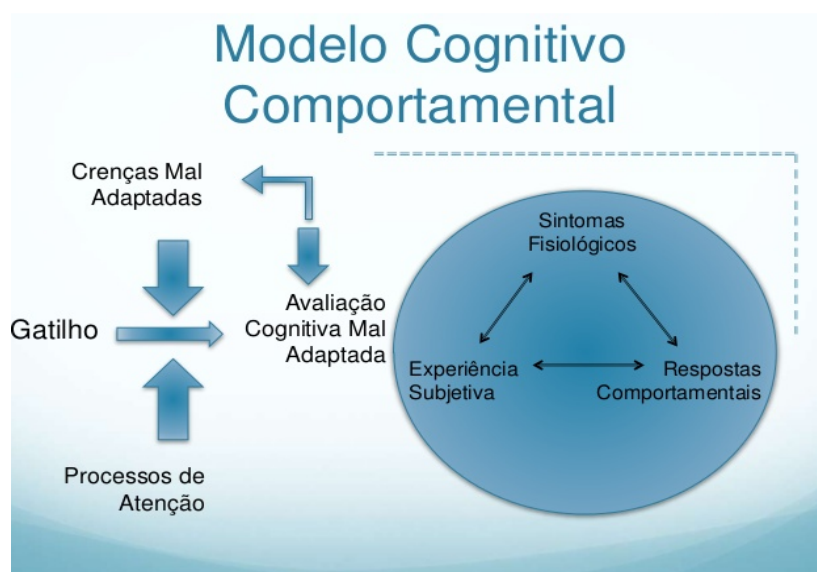
sentimentos e emoções na promoção de novos comportamentos dentro de uma visão mais ampla do mundo interno e o exterior.

No aconselhamento aos pais de crianças com DCC sobre o diagnóstico pré-natal, pode ser conceituado como o processo pelo qual os pais afetados passam por uma reação de luto, com seus conhecidos componentes de luto, choque, negação ou descrença, raiva, incluindo questionamento da experiência do clínico, culpa ou vergonha e tristeza com a sensação de estar sozinho. Há também um desejo ou "negociação" de um adiamento com a milagrosa perda do problema, seguida de uma eventual resolução e reconstrução (MENAHE, *et al.*, 2015).

Um dos primeiros objetivos do psicólogo cognitivo-comportamental no processo de aceitação de um diagnóstico é identificar as interpretações do paciente acerca de seu processo saúde-doença para iniciar um trabalho de reestruturação de pensamentos disfuncionais, adotando interpretações que tenham base em evidências existentes na realidade, ao invés da consideração de premissas irracionais (PEREIRA; PENIDO, 2010)

A maioria dos terapeutas cognitivos iniciam o tratamento do cliente ao identificarem os pensamentos automáticos e as distorções cognitivas frente à situação de crise e, em longo prazo, investigam as crenças intermediárias. Assim, a terapia cognitiva demonstra a importância do desenvolvimento da autonomia do paciente, treinando-o para que tenha habilidades para a modificação de pensamentos, comportamentos e/ou emoções disfuncionais (SOUSA *et al.*, 2015).

Figura 1 – Funcionamento Cognitivo



O papel do terapeuta, neste momento, é ajudar o paciente a pensar e agir de forma mais realista e adaptativa em relação aos seus problemas psicológicos e, deste modo, reduzir os sintomas (BECK *et al.*, 1997). Logo, cabe ao psicólogo analisar as variáveis que interferem na vida de cada paciente, para assim compreender como estas, influenciam em seus comportamentos e sentimentos frente à nova situação e possível adoecimento como depressão e ansiedade.

No estudo sobre o aconselhamento da psicologia da saúde e na relação clínica no aconselhamento, sugestão apresentada por Trindade e Teixeira (2000), componentes diferentes são apresentados, cujo peso específico pode variar em cada intervenção ou em cada entrevista em função das necessidades específicas do sujeito. Procuramos apresentar a sugestão de Trindade e Teixeira (2000) em formato de quadro para melhor didática (Quadro 2).

Quadro 2 – Componentes para intervenção clínica

Aconselhamento em psicologia da saúde	
Modelo	Intervenção
1	O aconselhamento ajuda na tomada das decisões, na mudança do comportamento para lidar com as dificuldades.
2	A psicoeducação ocorre através do conhecimento, que é transmitido das informações que precisam ser transmitidas para o paciente.
3	Através do aconselhamento, o paciente pode obter a segurança emocional, para o enfrentamento do tratamento, facilitação do controle interno e promoção da autonomia pessoal.

Fonte: Adaptado de Trindade e Teixeira (2000).

Alguns princípios gerais deste tipo de intervenção, nomeadamente: a elaboração de um plano de trabalho que inclui a compreensão do problema e a identificação das atividades necessárias para o superar; o treino de competências para aumentar a autoeficácia do sujeito no seu quotidiano; a atribuição da capacidade de mudança ao próprio sujeito; a utilização, pelo sujeito, das competências aprendidas fora do contexto de intervenção clínica. Scott e Dryden (1996), independentemente do modelo teórico utilizado, sugerem que o

aconselhamento deve seguir uma abordagem biopsicossocial (DAVY, 1999), ou seja, deve considerar, em simultâneo, o estado de saúde, o bem-estar psicológico, as competências sociais e a qualidade de vida do sujeito, promovendo a combinação da intervenção psicológica especializada com a intervenção médica (TRINDADE; TEIXEIRA, 2000). A história do sujeito, suas condições ambientais e experiências de modelagem pessoais devem ser consideradas, pois são tanto uma questão a ser elaborada terapeuticamente, como também sem dúvida a fonte da qual irá apresentar a solução para promover o equilíbrio psíquico emocional de enfrentamento à demanda ora manifesta.

O aconselhamento é complexo e devido à crescente relevância do componente humano no foco positivo da saúde em detrimento da doença. Assim, verificar os resultados do aconselhamento em saúde, identificar quem são os beneficiados direta e sistematicamente, pode indicar quais são os profissionais e por qual nível de preparo espera-se que tenham sob determinadas competências a serem desenvolvidas ou aperfeiçoadas.

Vale ressaltar que é importante a avaliação dos resultados do aconselhamento em saúde, entendendo-se que este tema é contemporâneo e a avaliação é complexa, uma vez que é necessário saber quem são os sujeitos que mais beneficiam com o aconselhamento em contextos de saúde e, também, qual o nível de competências de quem realiza a intervenção e que se relaciona com os benefícios do aconselhamento.

2.2 O DIAGNÓSTICO DA CARDIOPATIA CONGÊNITA NO FETO

A ultrassonografia pré-natal abriu novas oportunidades para examinar, diagnosticar e tratar o feto (HOWE, 2014). Assim, o aumento da demanda de casos de cardiopatia fetal e a crescente solução para diagnósticos acurados e prévios exigiram aperfeiçoamento de novas técnicas. Os exames de ultrassonografia obstétrica no segundo trimestre de gestação são procedimentos que deveriam ser oferecidos às gestantes com diversas finalidades médicas: calcular a idade gestacional, avaliar o número de fetos, localizar a placenta e rastrear anomalias fetais. Muitos países ao redor do mundo, incluindo a Suécia, implementaram esses procedimentos como parte dos cuidados de rotina da maternidade

(CRANG-SVALENIUS; DYKES; JÖRGENSEN, 1998). A grande maioria das gestantes aceita os exames oferecidos e a maioria acha fácil a realização desses exames (CARLSSON *et al.*, 2017). Um benefício importante do diagnóstico pré-natal precoce é que ele permite mais tempo para múltiplas oportunidades de revisar informações com os pais (LEE, 2017). A acuracidade e ampla utilização da ultrassonografia obstétrica tem permitido uma avaliação criteriosa do crescimento e bem-estar fetal (MENAHEM, 2012). De fato, as vantagens citadas anteriormente, seja em praticidade, precisão de diagnóstico, aderência e simpatia pelo exame, tem levado à promoção do diagnóstico intra útero de malformações congênitas, como a cardiopatia congênita fetal sendo as cardiopatias congênitas comuns em fetos, com uma incidência seis vezes maior do que as anormalidades cromossômicas (ROCHA *et al.*, 2014), é a causa mais comum de malformação congênita no feto (ROCHA *et al.*, 2013) que podem ensejar em níveis altos de morbidade significativa e mortalidade definida.

O diagnóstico pré-natal de uma anomalia fetal é um evento traumático na vida dos futuros pais (SANDELOWSK; BARROSO, 2005; WOOL, 2011), resultando em luto intenso e sofrimento psicológico (WOOL, 2011). Isso permitiu o aconselhamento dos pais afetados, que podem ter a opção de continuar ou não com a gravidez. (MENAHEM, 2012; CARLSSON *et al.*, 2017). Após o diagnóstico, os futuros pais expressam a necessidade de apoio e informações contínuas durante o restante da gravidez e têm preocupações com o futuro (BRATT *et al.*, 2015). De imediato, após o diagnóstico, a interrupção da gravidez parece ser o mais racional e espontâneo ato. Porém, o acesso à informação é essencial para promover um comportamento pensante que inclui planejar, julgar e resolver o conflito adotando medidas para tomada de decisão que venha a ser a mais adequada possível dentro do contexto biopsicossocial, podendo inclusive optar pela continuidade gestacional com o suporte clínico e psicológico ao longo dos próximos meses.

Particularmente na primeira visita, as reações iniciais de choque e tristeza a um achado anormal podem inibir a habilidade dos pais de reter informações (LEE, 2017). A consulta inicial é desafiadora, pois é necessário explicar as informações anatômicas e médicas potencialmente complexas a uma família quando os níveis de estresse são muito altos. Idealmente, haverá uma oportunidade de consulta(s) de acompanhamento para completar adequadamente o aconselhamento e reforçar

pontos que podem ter sido esquecidos em meio a um primeiro encontro emocional (LEE, 2017) Tal aconselhamento, entretanto, não é isento de dificuldades e dilemas éticos . Eles vão desde a tentativa de informar aos pais muito angustiados, a natureza e as implicações da DCC, o desfecho e os riscos envolvidos nas intervenções que podem ser necessárias, ao mesmo tempo em que é solicitado os desfechos em longo prazo. Tal aconselhamento, às vezes, baseia-se em informações incompletas obtidas ou se refere a lesões que podem evoluir durante o período gestacional. (MENAHEM, 2012). Os profissionais especialistas em cardiologia fetal têm o dever ético de manter seus conhecimentos teóricos e habilidades práticas para garantir que aconselham os pais corretamente (HOWE, 2014).

Além disso, as informações fornecidas não são capazes de levar em conta os possíveis avanços que podem ocorrer no futuro, que podem alterar a qualidade de vida e os resultados dos indivíduos afetados. Outros membros da família em questão podem desejar ter voz no processo de tomada de decisão. Os próprios clínicos podem querer levar em conta não apenas o fardo emocional da família e as possíveis intervenções como cirúrgicas ou não, internações, os riscos de complicações, etc., mas também os custos financeiros e outros suportados pela comunidade. (MENAHEM, 2012)

Diante do exposto, o diagnóstico pré-natal de DCC e seus benefícios são considerados um passo fundamental no manejo da cardiopatia congênita (RAKHA, 2020), requerendo do profissional de saúde habilidades e destrezas para além da constatação da cardiopatia, um acompanhamento contínuo, que garanta os desfechos acolhedores e necessários para cada caso.

2.3 JUSTIFICATIVA DA METODOLOGIA: REVISÃO DE ESCOPO

Um número crescente de pesquisas está agora colaborando com pessoas que vivenciam uma situação específica relacionada à saúde, como um diagnóstico pré-natal de malformação congênita (CARLSSON *et al.*, 2017). Pesquisadores que planejam colaborar com pessoas que têm experiência de diagnóstico pré-natal devem estar atentos às potenciais experiências emocionais associadas. A valorização relacionada ao encontro com os pares chama a atenção para a

necessidade de estudos que explorem o apoio dos pares. (CARLSSON *et al.*, 2017). Em todo o mundo, aos futuros pais deveriam ser oferecidos exame de ultra-som obstétrico, além da realização do ecocardiograma fetal, para monitorar a gravidez e avaliar a anatomia fetal. A introdução desses exames resultam em um aumento do número de anomalias fetais diagnosticadas no pré-natal, sendo as mais comuns as cardiopatias congênitas. (CARLSSON *et al.*, 2017). Dependendo das leis estaduais, no caso dos EUA e onde os abortos são permitidos de acordo com legislação em vigor, os futuros pais podem ter a opção de interromper a gravidez. A tomada de decisão de continuar ou interromper a gravidez envolve aspectos éticos, existenciais, psicológicos e médicos.

No entanto, há poucas pesquisas sobre a realização de aconselhamento pré-natal para DCC ou a determinação das estratégias mais eficazes para fornecer apoio familiar. É importante que os conselheiros tenham boas habilidades de comunicação, demonstrem empatia e sejam perspicazes na avaliação de como a informação está sendo recebida. O conselheiro deve avaliar a compreensão dos pais e o estado emocional durante a discussão (LEE, 2017). De fato, a decisão mediante este conflito é de natureza inesperada e com consequências ao longo do tempo que marcam a individualidade e a família envolvida pela questão e assim não se deve subtrair ou restringir este direito, pelo contrário, quando o paciente sente-se acolhido pode estar seguro para perceber-se como pessoa e adotar a melhor decisão com autonomia e responsabilidade consigo mesmo.

Perante o revelado, a assertividade do aconselhamento remete aos necessários, subsídios científicos com o objetivo de validar uma intervenção pautada nos achados que vislumbram a temática nos diversos aspectos que precisam ser considerados. Com o esforço de somar nesta construção, aqui foi escolhido à metodologia da revisão de escopo, por ser uma abordagem atual que, justamente por ter o objetivo inicial de analisar a pertinência do tema proposto, bem como descobrir a forma que se apresenta nos diversos bancos de dados científicos, se está consolidado ou não e em que ponto podemos considerar os achados.

3 MÉTODO

Como o aconselhamento familiar nos casos de cardiopatia congênita diagnosticada no período gestacional é um tema de extrema relevância e ainda emergente na literatura, a metodologia escolhida foi a síntese da revisão de escopo baseada nos princípios relatados por Fernández-Sánchez, King e Enríquez-Hernández (2020), preconizados pelo princípios de Joanna Briggs Institute e seguindo o checklist de revisão de escopo do PRISMA-ScR, Preferred Reporting Items for Systematic reviews and Meta-Analyses extension for Scoping Reviews (TRICCO *et al.*, 2018).

A revisão de escopo permite uma avaliação de novas literaturas sobre o tema, fornecendo bases significativas para pesquisas futuras. Como uma metodologia abrangente para examinar a literatura relevante em um determinado tema de pesquisa, as revisões de escopo têm o objetivo de examinar a extensão, o alcance e a natureza da atividade de pesquisa, bem como identificar lacunas de pesquisa na literatura existente.

Essa metodologia incluiu 5 etapas: identificação da questão da pesquisa; identificação dos estudos relevantes; seleção dos estudos; mapeamento dos dados e demonstração dos resultados. O protocolo dessa revisão de escopo foi registrado no Open Science Framework on 4 September 2022 (<https://doi.org/10.17605/OSF.IO/7WK45>).

3.1 ETAPA 1: IDENTIFICAÇÃO DA QUESTÃO DA PESQUISA

Considerando que existem poucos dados na literatura sobre aconselhamento dos familiares que recebem o diagnóstico de cardiopatia congênita no feto, e que ainda não estão resumidas ou sintetizadas, a questão da pesquisa foi: o que temos na literatura sobre o aconselhamento dos familiares que recebem diagnóstico de cardiopatia congênita no feto?

O acrônimo PCC (problema, conceito, contexto) foi o mnemônico que auxiliou na identificação dos tópicos-chave. Foi utilizado para nortear a coleta de dados estratégica e foi adotado para conduzir a questão de pesquisa (Quadro 3).

Quadro 3 – Descrição da estratégia PCC para formulação da pergunta de pesquisa

Acrônimo	Definição	Descrição
P	População	Familiares de fetos com cardiopatia congênita
C	Conceito	Aconselhamento familiar
C	Contexto	Período gestacional

Fonte: Cordeiro, S. A. (2021, p.21). Adaptado de: os autores (2022).

3.2 ETAPA 2: IDENTIFICAÇÃO DOS ESTUDOS RELEVANTES

Os estudos foram identificados por uma pesquisa sistemática realizada durante o mês de Setembro e Outubro de 2022. Foram utilizados 6 bancos de dados: Medline, Embase, Lilacs, Scielo, Scopus e Web of Science, além da literatura cinzenta: Psycinfo e Google acadêmico. Nosso intuito foi realizar uma busca completa: Medline e Embase foram escolhidas por serem os bancos de dados eletrônicos mais completos dentro da área da saúde, Lilacs reúne as revistas eletrônicas latino americanas, Scielo permite avaliar as revistas eletrônicas brasileiras, Scopus e Web of Science foram escolhidas por serem banco de dados mais abrangentes e se complementam. A literatura cinzenta foi incluída para ampliar o rastreo sobre as publicações relacionadas a este tema.

A estratégia de busca foi desenvolvida com a contribuição de um bibliotecário de pesquisa em ciências da saúde. Os descritores utilizados foram cardiopatia congênita, coração fetal e aconselhamento familiar. O resultado da estratégia de busca final para cada base de dados pode ser encontrada nos apêndices A, B, C, D, E, F, G, H, I, J e K. A tabela 1 demonstra os descritores utilizados para cada base de dados, com a utilização de filtro ou não, número de artigos e a data da busca mais recente. Todos os estudos identificados foram transferidos para o sistema Rayyan, sendo analisados de forma organizada e segura.

Tabela 1 – Descritores utilizados em cada base de dados

(continua)

Base de dados (portal)	Busca realizada	Filtro	Itens	Data da busca
-------------------------------	------------------------	---------------	--------------	----------------------

Medline (Pubmed)	((Counseling) AND ("Heart Defects, Congenital" [Mesh] OR "Defect, Congenital Heart" OR "Abnormality, Heart" OR "Heart Abnormality" OR "Congenital Heart Defect" OR "Heart, Malformation Of" OR "Malformation Of Heart" OR "Defects, Congenital Heart" OR "Heart Abnormalities" OR "Heart Defect, Congenital" OR "Congenital Heart Disease" OR "Congenital Heart Diseases" OR "Disease, Congenital Heart" OR "Heart Disease, Congenital" OR "Congenital Heart Defects") AND (fetal heart))	humanos	474	28/09/2022
Embase (Periódico Capes)	('counseling'/exp OR counseling OR 'parent counseling'/exp OR (counseling, AND parente) OR (guidance, AND parente) OR (parent AND guidance)) AND ('congenital heart malformation'/exp OR (congenital AND heart AND anomaly) OR (congenital AND heart AND defect) OR (congenital AND heart AND defects) OR (heart AND anomaly) OR (heart AND congenital AND anomaly) OR (heart AND congenital AND defect) OR (heart AND congenital AND malformation) OR (heart AND defects, AND congenital) OR (heart AND malformation)) AND ('fetus heart'/exp OR (fetal AND heart) OR (foetal AND heart) OR (heart, AND fetus)) AND [embase]/lim AND [humans]/lim	humanos	1188	28/09/2022
Lilacs (BVS)	(mh:"Aconselhamento" OR (consejo) OR (counseling) OR (conselho) OR mh: f02.784.176* OR mh:f04.408.413* OR mh:n02.421.143.303* OR mh:n02.421.461.363*) AND (mh: "Cardiopatias Congênitas" OR (cardiopatías congénitas) OR (heart defects, congenital) OR (malformação cardiovascular) OR (defeitos cardiovasculares congênitos) OR (anormalidades cardíacas) OR mh:c14.240.400* OR mh:c14.280.400* OR mh:c16.131.240.400*) AND (mh: "Coração Fetal" OR (corazón fetal))	título, resumos e assunto	19	28/09/2022

OR (fetal heart) OR
 mh:a07.541.278* OR
 mh:a16.378.303*) AND (
 db:("LILACS"))

Scielo	((Coração Fetal) OR (Corazón Fetal) OR (Fetal Heart)) AND ((Cardiopatias Congênicas) OR (Cardiopatías Congénitas) OR (Heart Defects, Congenital)) AND ((Aconselhamento) OR (Consejo) OR (Counseling))	sem filtro	7	01/10//2022
Scopus (Periódico Capes)	(ALL ("Hearts, Fetal" OR "Heart, Fetal" OR "Fetal Hearts") AND ALL ("Defect, Congenital Heart " OR "Abnormality, Heart Heart" OR "Abnormality Congenital Heart" OR "Defect Heart, Malformation Of" OR "Malformation Of Heart" OR "Malformation Of Hearts" OR "Defects, Congenital Heart" OR "Heart Abnormalities" OR "Heart Defect, Congenital" OR "Congenital Heart Disease" OR "Congenital Heart Diseases" OR "Disease, Congenital Heart" OR "Heart Disease, Congenital" OR "Congenital Heart Defects") AND ALL ("counseling" OR "counseling, parent" OR "parent counseling" OR "guidance, parent" OR "parent guidance"))	Medicine; Biochemistry, Genetics and Molecular Biology; Health Professions; Nursing; Multidisciplinary ; Psychology	577	01/10/2022
Web of Science	fetal heart and congenital heart disease and counseling	sem filtro	418	01/10/2022
Psycinfo (periódico Capes)	congenital heart disease AND counseling	sem filtro	46	01/10/2022
Google acadêmico	"fetal heart" and "congenital heart disease" and "counseling"	período de 2012 a 2022	primeiros 1010 artigos	01/10/2022

Fonte: Elaborada pela autora.

3.3 ETAPA 3: SELEÇÃO DOS ESTUDOS

Os critérios de inclusão foram:

- a) estudos sobre aconselhamento dos familiares que receberam o diagnóstico de cardiopatia congênita no feto;
- b) o aconselhamento familiar foi definido como qualquer profissional de saúde que realize orientação às mães e/ou aos pais sobre o diagnóstico de DCC durante o período gestacional; forma como a notícia é transmitida aos familiares, incluindo a explicação da DCC e questões sobre manejo e prognóstico;
- c) estudos empíricos e qualitativos, estudos quantitativos, sem data limite de publicação e qualquer idioma;
- d) excluímos da nossa revisão de escopo: os artigos em que não foi possível o texto completo;
- e) excluímos da nossa revisão: capítulos de livro, aulas, resumos publicados em anais de congresso, editoriais.

A primeira triagem realizada foi a exclusão de textos duplicados. Na sequência, tivemos a presença de 2 revisores trabalhando em pares, com a leitura dos títulos e resumos de todas as publicações, seguindo os critérios de elegibilidade. Após essa etapa, a leitura do texto completo foi realizada, concluindo assim a última fase de triagem dos estudos. Dúvidas e discordâncias foram resolvidas por consenso e discussão com um 3 revisor quando necessário. Foi realizada a contra referência (Anexo D) e a busca ativa de artigos sobre o tema.

3.4 ETAPA 4: MAPEAMENTO DOS DADOS

Um formulário de extração de dados foi desenvolvido no Excel e calibrado entre a equipe usando uma amostra randomizada. Os dados extraídos foram: autores, ano da publicação, localização do estudo, objetivos, metodologia, tamanho da amostra, equipe de aconselhamento e resultados importantes do estudo.

3.5 ETAPA 5: DEMONSTRAÇÃO DOS RESULTADOS

Os resultados desta revisão de escopo foram apresentados em narrativas, fluxograma, tabelas e quadros.

3.6 QUALIFICAÇÃO DOS REVISORES

Para a realização desta revisão, foram incluídos pesquisadores da área da saúde (psicologia e medicina):

- a) uma psicóloga (M.B.D.) formada pela Universidade Salesiana de São Paulo - UE de Lorena - UNISAL (2007) e aluna de mestrado da Pós Graduação em Ciências da Saúde (PPGCIS), da Faculdade de Medicina da Universidade Federal do Amazonas – UFAM;
- b) uma aluna (L.T.S.T.) do 8º período da Faculdade de Medicina da Universidade Federal do Amazonas – UFAM (2023);
- c) uma médica (L.A.R.A.) formada pela UFAM (2004), com especialização em cardiologia pediátrica e fetal pelo Instituto do Coração do Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo (InCor - HCFMUSP, 2009), com mestrado e doutorado em Ciências pela Universidade Federal de São Paulo (UNIFESP) e professora da PPGCIS - UFAM.

3.7 ASPECTOS ÉTICOS

Não existe conflito de interesse na realização deste estudo. Para revisões de literatura, não se faz necessário a análise e aprovação de um comitê de ética em pesquisa envolvendo os seres humanos.

4 RESULTADOS

Um total de 3719 artigos foram encontrados na busca inicial, considerando as bases de dados Medline (n = 474), Embase (n=1188), Scopus (n=557), Web of Science (n=418), Lilacs (n=19), Scielo (n=7) e na literatura cinzenta, Psycinfo (n=46) e google acadêmico (n=1010). Após a remoção de 1165 artigos duplicados e a exclusão de 2489 artigos apenas na leitura de títulos e resumos, permaneceram 65 artigos para a leitura do texto completo. Obedecendo os critérios de elegibilidade, 19 artigos permaneceram para análise. Foi realizada uma busca ativa e análise de contra referência, com o acréscimo de mais 2 artigos. Totalizando 21 artigos incluídos na revisão de escopo. A Figura 2 demonstra o fluxograma do estudo. A Tabela 2 resume os 21 artigos da revisão de escopo.

Figura 2 – Fluxograma da revisão de escopo.

(continua)

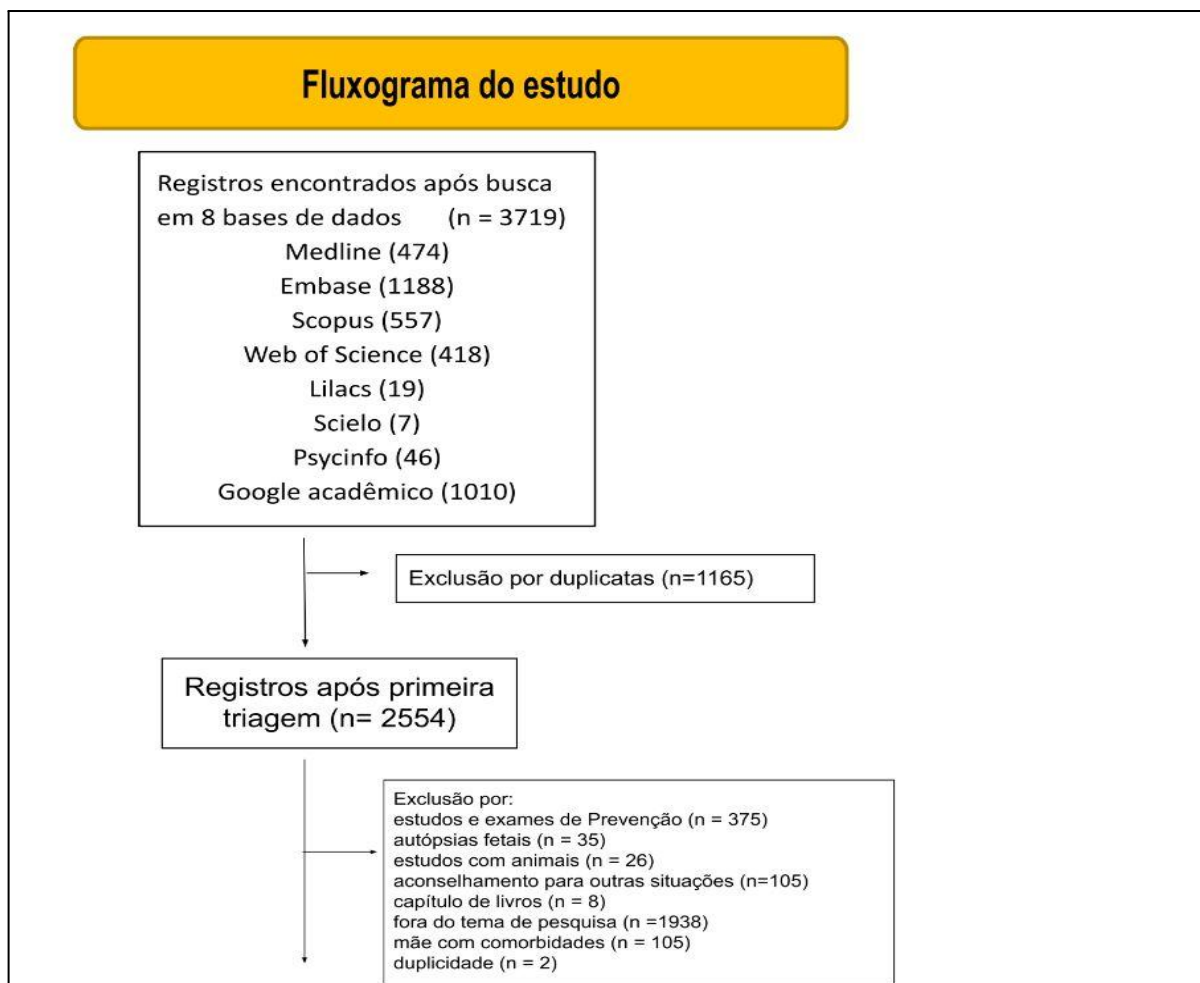
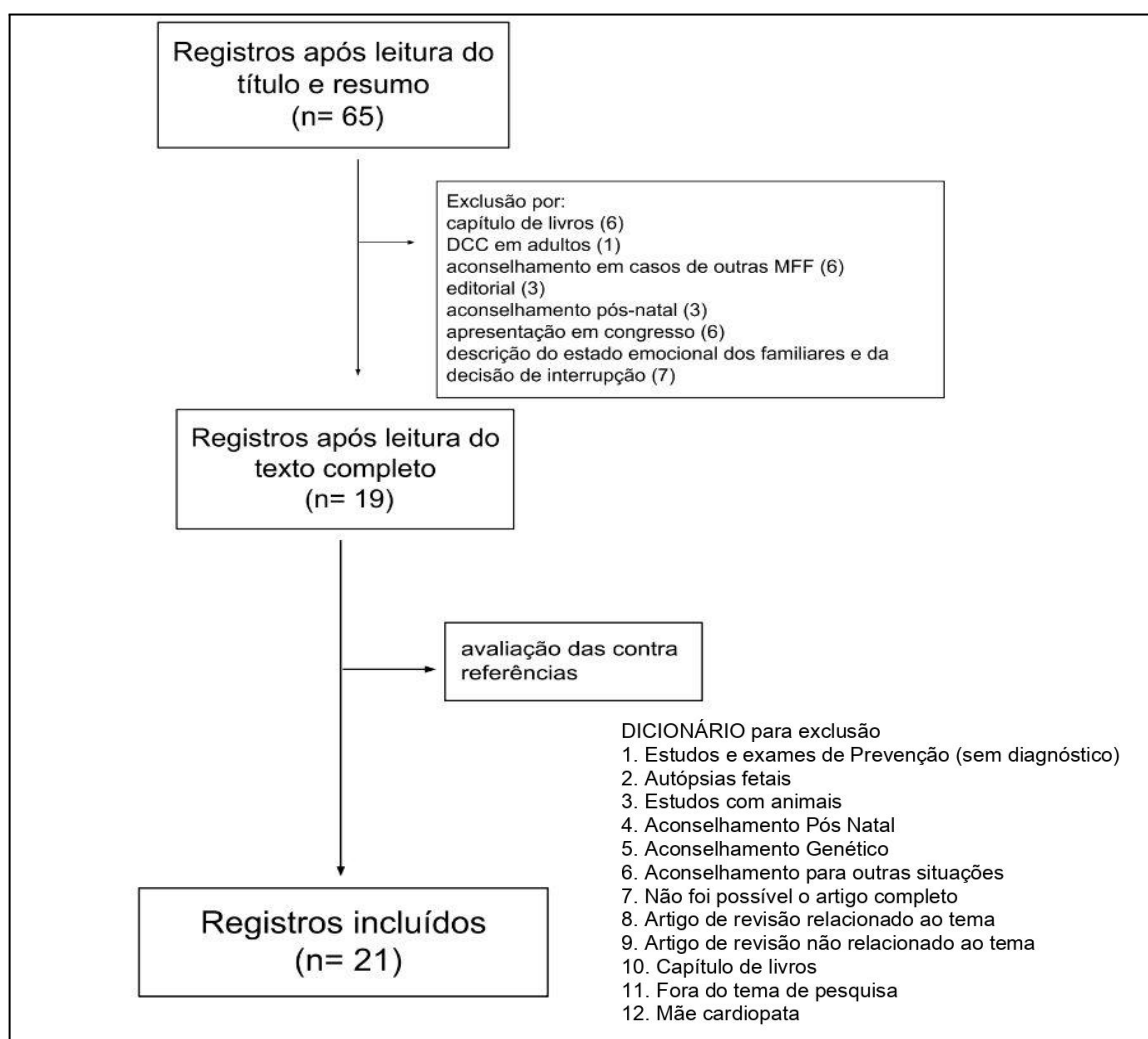


Figura 2 – Fluxograma da revisão de escopo.

(conclusão)



Fonte: Elaborado pela autora.

Tabela 2. Artigos da revisão de escopo sobre o aconselhamento familiar após o diagnóstico de doença cardíaca congênita no fetal

(continua)

Referências	Objetivo	Metodologia	Nº de participantes	Equipe de aconselhamento	Resultados
Menahem; Grimwade, Austrália 2004 (A1)	Avaliação dos familiares sobre o aconselhamento padronizado realizado no serviço.	Estudo qualitativo*	40 gestantes	CP, CF e MF. **	O aconselhamento melhorou a aceitação da CC e ajudou a entender o tratamento pós-natal, apesar de ser muito doloroso, melhor entendimento da CC com desenhos.

Tabela 2. Artigos da revisão de escopo sobre o aconselhamento familiar após o diagnóstico de doença cardíaca congênita no fetal

(continuação)

Referências	Objetivo	Metodologia	Nº de participantes	Equipe de aconselhamento	Resultados
Rempel, G.R. <i>et al.</i> Canadá. 2004 (A2)	Descobrir e descrever como os pais tomam decisões após o diagnóstico de DCC durante a gravidez e orientar os profissionais para o aconselhamento.	Estudo qualitativo	19 mães e 15 pais	CP,CF**	Os profissionais de saúde, incluindo enfermeiros, devem entender cada pai, estar cientes de sua influência profissional e apoiar a decisão de cada família.
Arya, B. <i>et al.</i> NY, EUA. 2013 (A3)	Determinar se o cardiologista e os familiares de crianças com DCC têm a mesma expectativa de informação e de aconselhamento entre o período pré-natal e pós-natal.	Estudo transversal	39 cardiologistas e 41 familiares	CP, CF	Os familiares acham que os cardiologistas devem aumentar a quantidade de informações relacionadas a DCC desde o pré-natal e continuar pelo resto da vida.
Hilton-Kamm, D; <i>et al.</i> Califórnia, EUA 2014 (A4)	Estudar as percepções e experiências dos pais ao receber o diagnóstico de DCC.	Estudo transversal *	841 respostas de pais de crianças com DCC	CP	A forma como as informações são apresentadas aos familiares podem moldar a tomada de decisão dos pais. Aplicação de questionário online com 82 perguntas.
Carlsson, T; <i>et al.</i> Suécia 2015 (A5)	Descrever as experiências dos pais após um diagnóstico pré-natal de DCC.	Estudo transversal *	6 pais e 5 mães de sete crianças com diagnóstico pré-natal de DCC.	CP	Valorização das informações precoces e honestas. As ilustrações como complemento às informações orais ajudam no entendimento da DCC. Fontes confiáveis na internet para maiores informações sobre a DCC.

Tabela 2. Artigos da revisão de escopo sobre o aconselhamento familiar após o diagnóstico de doença cardíaca congênita no fetal

(continuação)

Referências	Objetivo	Metodologia	Nº de participantes	Equipe de aconselhamento	Resultados
Bratt, E.L.; <i>et al.</i> Suécia, 2015 (A6)	Descrever sobre o aconselhamento nos casos de DCC no feto e as necessidades para um acompanhamento contínuo na gestação.	Estudo qualitativo*	6 casais	CP, CF, enfermeira, equipe obstétrica, psicólogo e assistente social. **	Curto prazo entre a suspeita e o diagnóstico preciso da DCC no feto, Aconselhamento contínuo na gestação. Informações escrita e de qualidade sobre a DCC. Explicações semelhantes para o parceiro. Contato contínuo com a enfermeira.
Carlsson, T, <i>et al.</i> Suécia 2016 (A7)	Descrever experiências dos familiares imigrantes suecos com diagnóstico de DCC no feto.	Estudo qualitativo*	5 gestantes e 4 parceiros masculinos	Enfermeira obstétrica	A necessidade de intérprete, informação visual, apoio psicossocial, respeitar a religião sobre a interrupção da gravidez
Carlsson, T.; <i>et al.</i> Suécia 2016 (A8)	Descrever as experiências dos pais após um diagnóstico pré-natal de DCC por meio de discussão em grupo.	Estudo qualitativo	3 mães e 2 pais em cada grupo	CP	Importante o suporte emocional do casal. Informações complementares sobre a DCC. Seguimento da gestante para repetir as informações sobre a DCC. Existiram 2 grupos: grupo que interrompeu e grupo que manteve a gestação
Walsh, M.J. <i>et al.</i> EUA 2017 (A9)	Avaliar o aconselhamento dos familiares com diagnóstico fetal de SHVE.	Estudo transversal*	201 participantes (57 familiares e 144 médicos)	CP, CF	Grande variabilidade entre as formas de aconselhamento. Dificuldade em abordar o prognóstico dessa doença. O desfecho precoce da doença é mais abordado do que o desfecho tardio. Aplicação de questionário aos cardiologistas e aos familiares.

Tabela 2. Artigos da revisão de escopo sobre o aconselhamento familiar após o diagnóstico de doença cardíaca congênita no fetal

(continuação)

Referências	Objetivo	Metodologia	Nº de participantes	Equipe de aconselhamento	Resultados
Im YM, <i>et al.</i> Coreia 2018 (A10)	Descrever experiência mães coreanas com diagnóstico pré-natal de DCC	Estudo qualitativo	12 mães	profissional de saúde especializada	Importância do aconselhamento familiar. Movimento do feto ajuda no entendimento de que o feto é um ser independente e que pode sofrer influências de fatores externos. Importância do aspecto religioso e espiritual (técnica de Taekyo)
Kovacevic, A; <i>et al.</i> Alemanha 2018 (A11)	Desenvolver um questionário adequado para avaliar as necessidades de aconselhamento dos pais.	Estudo transversal *	17 familiares	CP, MF e sociólogos.	O questionário aplicado pode ser uma ferramenta para avaliar o sucesso do aconselhamento familiar e recomenda um aconselhamento multidisciplinar.
Kovacevic, A; <i>et al.</i> Alemanha 2020 (A12)	Avaliação do aconselhamento familiar após o diagnóstico de DCC no feto.	Estudo transversal *	61 (40 mulheres e 21 homens)	CP, MF e sociólogos.	Aconselhamento familiar contínuo, com sala reservada, tempo de diálogo adequado, linguagem nativa, uso de informações escritas ou links para entendimento da CC. Sugere maior sucesso se realizado pelo cardiologista.
Kovacevic, A.; <i>et al.</i> Alemanha ,2020 (A13)	Avaliar o aconselhamento fornecido aos familiares com diagnóstico de DCC no feto.	Estudo transversal multicêntrico*	168 familiares	CP,CF e MF	Transparência das informações e do processo de tratamento melhoram o aconselhamento. DCC mais complexas são melhores esclarecidas pelos cardiologistas pediátricos

Tabela 2. Artigos da revisão de escopo sobre o aconselhamento familiar após o diagnóstico de doença cardíaca congênita no fetal

(continuação)

Referências	Objetivo	Metodologia	Nº de participantes	Equipe de aconselhamento	Resultados
Bertaud S.; <i>et al.</i> Inglaterra 2020 (A14)	Avaliar o aconselhamento dos familiares com diagnóstico fetal de SHVE.	Estudo qualitativo*	8 mães	CP, CF	O aconselhamento desses familiares oferecem uma melhor visão das perspectivas futuras do seu filho.
Delaney, R.K.; <i>et al.</i> EUA.2021 (A15)	Avaliar o efeito de 2 protocolos de aconselhamento familiar em casos de DCC no feto.	Ensaio clínico randomizado	35 familiares	CP, CF, cirurgião, assistente social, paliativistas	Publicação do protocolo do estudo
Holmes, K. W. <i>et al.</i> Portland, EUA 2022 (A16)	Avaliar a compreensão dos familiares sobre os defeitos cardíacos fetais após o aconselhamento.	Estudo transversal *	39	CP, CF e enfermeira	O aconselhamento fetal foi eficaz para transmitir a anatomia e a necessidade de cirurgia. Houve menor compreensão para mulheres com menos instrução. Foram avaliados: descrição da condição cardíaca; quão confiantes estavam no diagnóstico e se o feto precisaria de cirurgia cardíaca.
Kovacevic, A; <i>et al.</i> Alemanha 2021 (A17)	Avaliar o aconselhamento familiar para DCC fetal durante e antes da pandemia do COVID-19	Estudo transversal multicêntrico*	226 familiares	CP,CF e MF	Não houve diferença significativa entre os grupos em relação ao sucesso do aconselhamento desses familiares mesmo com a pandemia do COVID-19.

Tabela 2. Artigos da revisão de escopo sobre o aconselhamento familiar após o diagnóstico de doença cardíaca congênita no fetal

(conclusão)

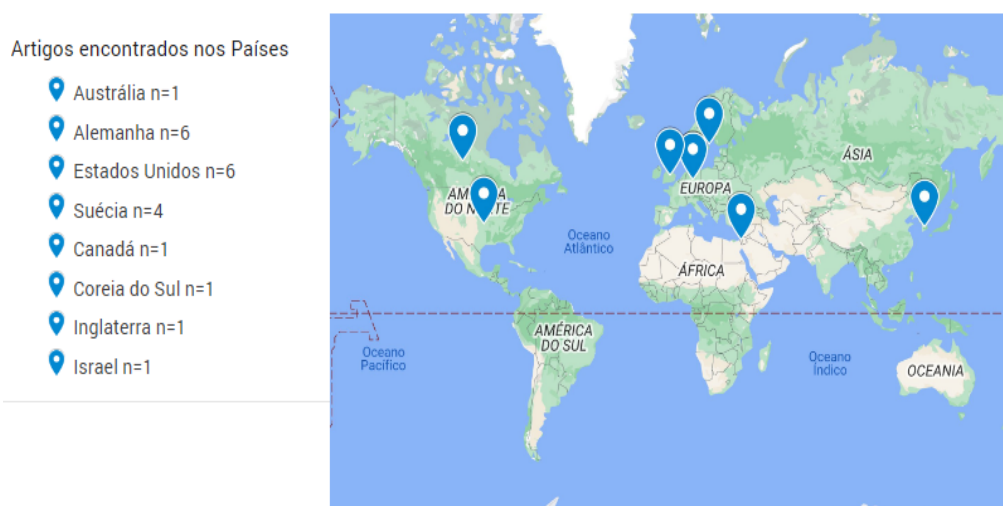
Referências	Objetivo	Metodologia	Nº de participantes	Equipe de aconselhamento	Resultados
Gendler, Y; <i>et al.</i> Israel 2021 (A18)	Avaliação dos familiares sobre o aconselhamento padronizado realizado no serviço.	Estudo transversal *	100 casais	CP	O aconselhamento padronizado usando um checklist auxilia no fornecimento de informações. Satisfação dos pais no processo de aconselhamento. Boa percepção dos cardiologistas em relação ao entendimento da DCC pela família. Aplicação de 2 questionários: um para o CP e outro para os pais.
Kovacevic, A; <i>et al.</i> Alemanha 2021 (A19)	Fornecer uma visão geral atualizada da literatura avaliando o aconselhamento em relação às necessidades dos pais.	Revisão integrativa	Não se aplica.	Não se aplica.	Proposta de como otimizar um cenário de aconselhamento com base na literatura atual e na experiência do serviço.
Harris, K. W. <i>et al.</i> Pittsburgh, EUA 2022 (A20)	Descrever a experiência dos pais que recebem o diagnóstico de DCC fetal (pré e pós-natal)	Estudo qualitativo*	17 famílias	CP	Incerteza do diagnóstico e a falta de informação adequada aumenta o estresse das famílias. Familiares preferem os cardiologistas fetais para o esclarecimento e enfrentamento da situação.
Kovacevic, A; <i>et al.</i> Alemanha. 2022 (A21)	Identificar quais fatores desempenham um papel importante para o sucesso do aconselhamento familiar nos casos de DCC no feto.	Estudo transversal multicêntrico*.	226	CP,CF e MF	Curto tempo entre a suspeita e o diagnóstico preciso da DCC e a explicação pelo especialista. Informação sobre a DCC de forma clara e ilustrada. Apoio pelo médico que aconselhou. Segurança no local de tratamento.

Fonte: Elaborado pela autora.

CP: cardiologista pediátrico, CF: cardiologista fetal, MF: médico fetal. * Aplicação de um questionário como ferramenta de avaliação da efetividade do aconselhamento familiar. **Geneticista, pediatra e cirurgião pediátrico (se necessário).

Foram incluídos 21 artigos, todos realizados em países desenvolvidos (Figura 3) e nos últimos 10 anos foram publicados 19 (A3 até A21) dos 21 artigos sobre o tema. As pesquisas foram realizadas com metodologia qualitativa em oito artigos, (A1, A2, A6, A7, A8, A10, A14, A20), uma revisão integrativa (A19) um protocolo de ensaio clínico randomizado (A15) e os demais foram estudos transversais, (A3, A4, A5, A9, A11, A12, A16, A18), sendo 3 multicêntricos (A13, A17, A21). Observamos que 2 artigos avaliaram o aconselhamento familiar em uma cardiopatia específica: a síndrome de hipoplasia do ventrículo esquerdo (SHVE) (A19, A14).

Figura 3 – Mapa demonstrando os países onde os estudos foram realizados.



Fonte: Elaborada pela autora. Disponível em: <https://www.google.com/maps/d/u/0/edit?mid=18XlzkANshZZbcAP3d77QP_kPCEL_F_4&ll=49.030730356991185%2C0&z=2>.

Todos os artigos incluídos buscam compreender as percepções e experiências dos pais ao receber o aconselhamento após o diagnóstico de DCC no período gestacional. Entretanto, A9 e A18 foram os únicos que consideraram o ponto de vista do profissional de saúde na avaliação do aconselhamento familiar. Nessas duas publicações, os profissionais de saúde relatam que o familiar compreende a cardiopatia, no entanto, ao avaliar o questionário dos familiares se percebe ainda uma dificuldade na compreensão da doença. Existem ainda situações em que o profissional de saúde apresenta algumas dificuldades em esclarecer sobre

o prognóstico de cardiopatias mais complexas com alterações mais importantes, como demonstrado em A9. Apesar do diagnóstico ser muito doloroso, o aconselhamento consegue melhorar a aceitação da DCC e ajuda na compreensão da doença, do tratamento e do prognóstico, segundo A1.

Não foi encontrado nenhum estudo que avaliasse diretamente sobre qual profissional tinha melhor resultado no aconselhamento desses familiares. Porém, pelo menos 5 estudos (A3, A12, A13, A18, A20) descreveram que a conversa com um especialista em cardiologia pediátrica e fetal era mais detalhada, deixando os familiares mais seguros e tranquilos com as informações, evidenciando a importância da presença do cardiologista pediátrico no diagnóstico e no aconselhamento familiar. Apenas um estudo (A6) abordou especificamente sobre o aconselhamento com a presença de um psicólogo de saúde na equipe.

Observamos uma tendência, dos serviços de cardiologia fetal, em realizarem questionários para avaliação do grau de satisfação dos familiares no aconselhamento recebido durante o período gestacional. Todos os estudos transversais e qualitativos utilizaram algum questionário com intuito de avaliar a efetividade do aconselhamento familiar, sendo considerados alguns desses critérios: a adequada compreensão dos pais em relação ao diagnóstico da DCC, o apoio do médico diante deste diagnóstico, facilidade na compreensão da DCC, duração do tempo entre a suspeita da DCC e a consulta com o especialista (cardiologista fetal) e o aconselhamento, a capacidade de atendimento médico adequado para receber o bebê com DCC, informações sobre o prognóstico do bebê, tipo de tratamento necessário para cada situação, o tempo em que esse tratamento fosse necessário acontecer e o processo de tomada de decisão para a interrupção da gestação (A18). Dentre os artigos, pelo menos 6 relataram sobre a importância da utilização de desenhos e ilustrações por sites seguros como facilitador na compreensão da doença.

O aconselhamento familiar para DCC fetal antes e durante a pandemia de COVID-19 foi analisado pelo grupo alemão (A17), que demonstrou não haver qualquer diferença na eficácia do processo; no entanto, os pais aconselhados durante a pandemia estavam significativamente mais preocupados.

Observou-se ainda que a relação entre o nível intelectual das mães estava diretamente relacionado com a forma de compreensão sobre o aconselhamento na

transmissão da anatomia e da necessidade de cirurgia (A16).

A percepção do feto como um ser independente que pode receber influências externas, muito relacionado a aspectos religiosos e espirituais, foi o destaque de A10. Nesse artigo, os familiares utilizam a técnica de Taekyo para auxiliar na compreensão e na aceitação diante de um diagnóstico inesperado de DCC no feto (A10).

6 DISCUSSÃO

O conhecimento dos mecanismos fisiopatológicos das cardiopatias congênitas é necessário para que se possa acompanhar a progressão da doença, desde o diagnóstico até as possíveis complicações futuras. Desse modo, o aconselhamento familiar ainda na fase pré-natal é de suma importância no processo de entendimento dos genitores, possibilitando uma abordagem eficaz que apresenta a realidade de forma didática e transparente (LEBOVICI, 2004).

O aconselhamento, além de estabelecer um canal de comunicação entre o profissional e os pais para as tomadas de decisões terapêuticas e informar o prognóstico, é uma ferramenta para o restabelecimento do equilíbrio emocional e da crise individual e familiar (GOMES *et al.*, 2010). Dessa maneira, a qualidade do aconselhamento familiar é influenciada principalmente pelo tempo gestacional, sendo preferencialmente no primeiro trimestre da gestação, pela equipe multiprofissional responsável pelo acompanhamento da gestante e a forma em que as informações e o apoio emocional são transmitidos.

Nesta revisão de escopo, notou-se que as dificuldades em comum apresentadas nos estudos foram: a barreira linguística, sociocultural e educacional entre o profissional do aconselhamento e a família (A7, A16). A complexidade das informações acerca da cardiopatia congênita torna-se um obstáculo para a compreensão dos pais na hora do aconselhamento, o que leva a perspectivas irreais dos genitores sobre a qualidade de vida dos filhos e os prognósticos terapêuticos.

Pode-se afirmar ainda a maior importância da presença paterna no aconselhamento, pois apenas 4 estudos (A1, A10, A14 e A19) focaram apenas na experiência das gestantes. Portanto, a presença familiar é de suma importância para a qualidade do serviço.

Outro obstáculo é qual profissional deve realizar o aconselhamento; não há evidências, a partir dos estudos, de formas avaliativas da efetividade do aconselhamento conforme cada profissional. Entretanto, como a maioria das pesquisas - A1, A2, A3, A4, A5, A6, A8, A9, A10, A11, A12, A13, A14, A15, A16 A17, A18, A20, A21 - havia a presença de um cardiologista pediátrico, pode-se inferir a necessidade de um profissional especializado para sanar as dúvidas e transmitir segurança aos pacientes. A falta de um apoio multidisciplinar e a falta de técnicas de

aconselhamento podem tornar ainda mais difícil a compressão sobre o diagnóstico e o tratamento da cardiopatia.

Apesar do aconselhamento gerar melhores resultados na saúde emocional familiar, há uma grande escassez de pesquisas com alta evidência científica, já que nesta revisão apenas um protocolo de ensaio clínico randomizado (A15), que está em andamento, foi identificado. Dessa forma, como os estudos A3, A4, A5, A9, A11, A12, A16 e A18 são majoritariamente de metodologia transversal, a noção de temporalidade é inexistente, o que impossibilita analisar os impactos da continuidade do aconselhamento, uma queixa repetidamente dita pelos familiares.

Outra característica observada nesta revisão de escopo é a recente publicação dos estudos, dos 21 artigos, A3, A4, A5, A6, A7, A8 A9, A10, A11, A12, A13, A14, A15, A16, A17, A18, A19, A20, A21 são dos últimos 5 anos, ou seja, análises retrospectivas dos serviços de aconselhamento familiar são escassos, conseqüentemente não há maneiras de avaliar melhorias ou obstáculos durante os anos. Outrossim, como o local da realização das pesquisas são em países desenvolvidos, ainda não há dados sobre o impacto e o funcionamento do aconselhamento em países em desenvolvimento, como o Brasil. Dessa forma, perpetua-se a falta de dados acerca da influência sociocultural das gestantes, como escolaridade, religião e renda, no nível de compreensão das DCC e nas tomadas de decisões terapêuticas.

No decorrer da leitura dos estudos dessa revisão, constatou-se que não existe metodologia sistematizada para o aconselhamento descrito na literatura. Ou seja, como não há dados suficientes para a validação de métodos de avaliação da compressão dos familiares e do impacto do aconselhamento, perpetua-se diversas lacunas acerca do serviço, principalmente a insistência de programas de capacitação para os profissionais de saúde. Dessa forma, o aconselhamento familiar é influenciado pela estrutura dos locais de atendimento, variáveis socioculturais e pela subjetividade dos parâmetros utilizados.

Em um estudo qualitativo e longitudinal foi possível observar que os pais valorizavam as informações que eram passadas de forma clara, objetiva e realista (A21). A Associação Europeia de Cardiologia Pediátrica e Congênita (AEPC) orienta que sejam usados diagramas, desenhos manuais, modelos ou instrumentos online para facilitar o entendimento dos pais, entretanto, devido escassez desses métodos,

fica inviável facilitar a adesão, pois os materiais disponíveis ainda não possuem validação e os conteúdos online em sua maioria não são confiáveis (MAGALHÃES *et al.*, 2016). Diante disto, torna-se necessário realizar um estudo de caráter metodológico utilizando a tecnologia, sendo uma ferramenta a mais para promover informações fidedignas sobre as cardiopatias congênitas.

Ademais, durante a busca por literatura, há forte tendência de estudos que avaliam a situação emocional dos familiares em relação ao diagnóstico de DCC nos serviços de saúde (A8). Tais estudos não foram selecionados para essa revisão de escopo por estarem fora do tema. Esses estudos descreviam sobre o grau de depressão e ansiedade dos familiares diante do diagnóstico, durante o período gestacional e como esse estado emocional poderia influenciar a relação entre os pais e o recém-nascido com DCC.

Observa-se uma maior preocupação dos pesquisadores em avaliar se o aconselhamento está sendo realmente efetivo para os familiares, quais os pontos fortes e quais os pontos fracos desse aconselhamento para serem melhorados na instituição. Essas avaliações estão sendo realizadas por meio de questionários na grande maioria dos estudos: A4, A9, A11, A12, A13, A14, A15, A16, A17 e A18. Os questionários são aplicados aos familiares e/ou aos profissionais de saúde conselheiros e realizados por entrevistas pessoalmente, pela Internet ou por telefone, apenas um estudo específico realizou uma discussão entre grupo.

6.1 PONTOS FORTES DO ESTUDO

Esta é a primeira revisão de escopo sobre este tema, desta forma, foi possível identificar diversas lacunas sobre esse tema tão relevante.

6.2 LIMITAÇÕES DO ESTUDO

A qualidade dos estudos selecionados não foi avaliada, o que não é obrigatório para este tipo de revisão.

6.3 QUESTÕES PARA NOVAS PESQUISAS

Evidenciamos que os estudos de aconselhamento foram realizados exclusivamente em países desenvolvidos, o que gera questionamentos sobre esta ferramenta em uma população com recursos escassos. Desde avaliações qualitativas em diferentes subpopulações até estudos de coorte avaliando o tempo de aconselhamento, número de consultas e interação sobre o estresse de sobrecarga psicológica da família podem ser melhor estudados. Ainda, podem ser realizados estudos validando os questionários utilizados para se identificar o grau de compreensão dos pais sobre a doença. Assim como se identificar se existe diferença deste grau de compreensão se a informação for passada por um cardiologista pediátrico ou um médico fetal ou ultrassonografista.

7 CONCLUSÃO

O presente estudo identificou que as evidências apontam que o aconselhamento familiar auxilia no processo de compreensão da cardiopatia congênita do feto. A maioria dos estudos são qualitativos e de desenho transversal, mas, não se sabe até que ponto ele pode amenizar o impacto psicológico da notícia. Ainda, existem questionários que podem ser utilizados para se identificar o grau de compreensão do familiar. Os fatores que mais parecem impactar nessa compreensão são a escolaridade, tempo de duração da explicação sobre a DCC e utilização de diagramas e sites sobre a DCC.

Não identificamos estudos que apontem se existe influência no processo de compreensão da cardiopatia congênita do feto em relação ao tipo de profissional de saúde que realizou o aconselhamento familiar.

De acordo com os estudos a uma necessidade de ampliar e fortalecer as condutas de aconselhamento fetal, principalmente sobre qual caminho devem seguir, qualidade de vida e prognóstico do feto, para que a família possa se preparar para os próximos passos, tais como, a escolha onde fazer o seguimento, providenciar o parto em um ambiente apropriado e obter mais informações referente a cardiopatia. Neste contexto, a criação de métodos/pesquisas ou validação de questionários é muito importante e tem grande potencial para mudar a realidade dos prognósticos de Dcc no Brasil. Os achados evidenciaram a relevância do tema proposto, uma vez que a necessidade do aconselhamento aos familiares após o diagnóstico de DCC no feto deve ser contínua e deve abarcar diferentes e importantes pontos, desde o diagnóstico e o entendimento da doença aos fatores biopsicossociais.

Conseguimos caracterizar os desenhos de estudo disponíveis na literatura sobre o aconselhamento dos familiares que receberam o diagnóstico de cardiopatia congênita fetal. Apesar de ser uma temática mundial de longa data, os achados foram recentes, e os estudos preconizados por países desenvolvidos, as metodologias vão desde uma revisão integrativa, apresentando o cenário de aconselhamento com base na literatura atual e na experiência do serviço, Identificamos ferramentas que avaliam o aconselhamento familiar utilizado para os casos de cardiopatia congênita no feto nos estudos transversais com aplicação de

entrevistas e questionários. Também identificamos estudo de ensaio clínico randomizado comparando efeito de dois protocolos de aconselhamento.

Embora o aconselhamento auxilie a compreensão da DCC, não há evidências fortes que relacionem a implementação com menores escores de sofrimento psíquico materno e familiar ou na decisão terapêutica ou de interrupção gestacional.

A compreensão das cardiopatias congênitas continua sendo um desafio para os profissionais da área, tanto pela pouca capacitação do tema na graduação em saúde, quanto pela presença de fatores inerentes que impactam no aconselhamento, como escolaridade materna, tempo gestacional e estrutura do ambiente de atendimento.

Observamos que existem lacunas neste assunto, norteadas melhor o que devemos estudar, como por exemplo, a avaliação sobre a interação entre o processo de aconselhamento familiar e a decisão de interrupção da gestação aos preceitos éticos e que tipo de problema e necessidades os familiares e os profissionais que transmitem essa notícia apresentam, se o tipo de profissional que orientou os familiares impactou nesta compreensão e ainda se existe o acompanhamento em psicoterapia individual e grupal.

Por fim, os resultados desta revisão demonstraram que existem poucos dados na literatura sobre aconselhamento dos familiares que recebem o diagnóstico de cardiopatia congênita no feto, e que ainda não estão resumidas ou sintetizadas sobre o aconselhamento dos familiares que recebem diagnóstico de cardiopatia congênita no feto. Pesquisas futuras são fundamentais para ampliar e fortalecer as condutas de aconselhamento fetal.

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APÊNDICE A - RESULTADOS DE BUSCA NO PUBMED

PMID	Title	Authors	Citation / DOI	First Author	Journal/ Book	Publication Year	Create Date	PMCID	NIHMS ID
35930815	Assisted reproduction in patients with cardiac disease: A retrospective review	Skorupskaite K, Joy E, Balen A, Agarwal K, Cauldwell M, English K.	Eur J Obstet Gynecol Reprod Biol. 2022 Sep;276:19-203. doi: 10.1016/j.ejogrb.2022.07.020. Epub 2022 Jul 29.	Skorupskaite K	Eur J Obstet Gynecol Reprod Biol	2022	2022/08/05		
35524915	Cardio-Obstetrics: the Next Frontier in Cardiovascular Disease Prevention	Thakkar A, Hailu T, Blumenthal RS, Martin SS, Harrington CM, Yeh DD, French KA, Sharma G.	Curr Atheroscler Rep. 2022 Jul;24(7):493-507. doi: 10.1007/s11883-022-01026-6. Epub 2022 May 7.	Thakkar A	Curr Atheroscler Rep	2022	2022/05/07	PMC9076812	
35507001	Transition in cardiology 2: Maternal and fetal congenital heart disease	Nagata H, Yamamura K, Matsuoka R, Kato K, Ohgas S.	Pediatr Int. 2022 Jan;64(1):e15098. doi: 10.1111/ped.15098.	Nagata H	Pediatr Int	2022	2022/05/04		
35490924	Reproductive Issues in Patients With the Fontan Operation	Wichert-Schmitt B, D'Souza R, Silversides CK.	Can J Cardiol. 2022 Jul;38(7):921-929. doi: 10.1016/j.cjca.2022.04.020. Epub 2022 Apr 28.	Wichert-Schmitt B	Can J Cardiol	2022	2022/05/01		
35484660	[Follow-up study of fetal cardiac birth defects after prenatal diagnosis and	Shen JJ, Pang CC, Yang LQ, Lin XY, Wang YY, Huang YP, Li YF, Pan W.	Zhonghua Fu Chan Ke Za Zhi. 2022 Apr 25;57(4):278-283. doi:	Shen JJ	Zhonghua Fu Chan Ke Za Zhi	2022	2022/04/29		

	graded counseling]		10.3760/cm a.j.cn11214 1-20211103-00639.						
35482 990	A prenatal case of Simpson-Golabi-Behmel syndrome type 1 with a 0.26-Mb deletion fragment at Xq26.2 inherited from mother: Case report	Sha J, Tan F, Liu Y, Xu Z, Wang X, Zhai J.	Medicine (Baltimore). 2022 Apr 22;101(16):e29222. doi: 10.1097/MD.00000000000029222.	Sha J	Medicine (Baltimore)	2022	2022/04/28	PMC9276221	
35429 589	Prenatal Diagnosis and Management of Single-Ventricle Heart Disease	Freud LR, Seed M.	Can J Cardiol. 2022 Jul;38(7):897-908. doi: 10.1016/j.cjca.2022.04.003. Epub 2022 Apr 13.	Freud LR	Can J Cardiol	2022	2022/04/16		
35278 231	Being born with a single cardiac ventricle: What do we tell prospective parents	d'Udekem Y, Hutchinson D.	Prenat Diagn. 2022 Apr;42(4):411-418. doi: 10.1002/pd.6121. Epub 2022 Apr 7.	d'Udekem Y	Prenat Diagn	2022	2022/03/12		
35262 959	Motion corrected fetal body magnetic resonance imaging provides reliable 3D lung volumes in normal and abnormal fetuses	Davidson J, Uus A, Egloff A, van Poppel M, Matthew J, Steinweg J, Deprez M, Aertsen M, Deprest J, Rutherford M.	Prenat Diagn. 2022 May;42(5):628-635. doi: 10.1002/pd.6129. Epub 2022 Mar 15.	Davidson J	Prenat Diagn	2022	2022/03/09	PMC9310761	

35238 062	Prenatal diagnosis of congenital ventricular aneurysm and diverticulum: Prenatal features and perinatal management	Morin C, Ponzio A, Guirgis M, Benzouid C, Beyler C, Rosenblatt J.	Prenat Diagn. 2022 Apr;42(4):4 28-434. doi: 10.1002/pd.6122. Epub 2022 Mar 8.	Morin C	Prenat Diagn	2022	2022/03/03		
35210 008	Counseling for Perinatal Outcomes in Women with Congenital Heart Disease	Gerardin JF, Cohen S.	Clin Perinatol. 2022 Mar;49(1):4 3-53. doi: 10.1016/j.clp.2021.11.004. Epub 2022 Jan 21.	Gerardin JF	Clin Perinatol	2022	2022/02/25		
34981 204	Prenatal diagnosis and postnatal outcomes of right aortic arch anomalies	Topbas Selcuki NF, Senol G, Esin D, Ozkose ZG, Caypinar SS, Bornaun H, Cetin BA, Yuksel MA.	Arch Gynecol Obstet. 2022 Sep;306(3): 745-752. doi: 10.1007/s00404-021-06346-7. Epub 2022 Jan 4.	Topbas Selcuki NF	Arch Gynecol Obstet	2022	2022/01/04		
34952 671	Outcome and Impact of Associated Left-Sided Cardiac Lesions in Coarctation of the Aorta Diagnosed During Fetal Life	Polyviou S, Charakida M, Miller OI, Rscn TW, Vigneswaran TV, Zidere V, Day TG, Lloyd DFA, Sharland GK, Simpson JM.	Am J Cardiol. 2022 Mar 1;166:114-121. doi: 10.1016/j.amjcard.2021.11.023. Epub 2021 Dec 21.	Polyviou S	Am J Cardiol	2022	2021/12/25		
34842 935	Contribution of fetal magnetic resonance imaging in fetuses with	Goncalves LF, Lindblade CL, Cornejo P, Patel MC, McLaughlin	Pediatr Radiol. 2022 Mar;52(3):5 13-526. doi:	Goncalves LF	Pediatr Radiol	2022	2021/11/29		

	congenital heart disease	ES, Bardo DME.	10.1007/s00247-021-05234-1. Epub 2021 Nov 29.						
34689 682	Presence of Cervical Vertebral Anomalies with Concomitant Non-Communicating Hydrocephalus and Multicystic Kidney in a Female Fetus: Where VACTERL-H Meets MURCS	Dracopoulos C, Gembicki M, Scharf JL, Welp A, Berg N, Weichert J.	Fetal Pediatr Pathol. 2022 Oct;41(5):871-880. doi: 10.1080/15513815.2021.1994068. Epub 2021 Oct 23.	Dracopoulos C	Fetal Pediatr Pathol	2022	2021/10/25		
34680 874	Adverse Perinatal and Early Life Outcomes following 15q11.2 CNV Diagnosis	Chu FC, Shaw SW, Lee CH, Lo LM, Hsu JJ, Hung TH.	Genes (Basel). 2021 Sep 23;12(10):1480. doi: 10.3390/genes12101480.	Chu FC	Genes (Basel)	2021	2021/10/23	PMC8535766	
34510 635	Genetic counseling for congenital heart disease - Practice resource of the National Society of Genetic Counselors	Ison HE, Griffin EL, Parrott A, Shikany AR, Meyers L, Thomas MJ, Syverson E, Demo EM, Fitzgerald KK, Fitzgerald-Butt S, Ziegler KL, Schartman AF, Stone KM, Helm BM.	J Genet Couns. 2022 Feb;31(1):9-33. doi: 10.1002/jgc.4.1498. Epub 2021 Sep 12.	Ison HE	J Genet Couns	2022	2021/09/12		
34405 872	Management of acute cardiovascular	Egidy Assenza G, Dimopoulos K, Budts W, Donti A,	Eur Heart J. 2021 Nov 1;42(41):4224-4240.	Egidy Assenza G	Eur Heart J	2021	2021/08/18		

	complications in pregnancy	Economy KE, Gargiulo GD, Gatzoulis M, Landzberg MJ, Valente AM, Roos-Hesselin k J.	doi: 10.1093/eurheartj/ehab546.						
34331895	Association between pregnancy and long-term cardiac outcomes in individuals with congenital heart disease	Son SL, Hosek LL, Stein MC, Allshouse AA, Catino AB, Hoskoppal AK, Cox DA, Whitehead KJ, Lindsay IM, Esplin S, Metz TD.	Am J Obstet Gynecol. 2022 Jan;226(1):124.e1-124.e8. doi: 10.1016/j.ajog.2021.07.015. Epub 2021 Jul 28.	Son SL	Am J Obstet Gynecol	2022	2021/07/31	PMC8748281	NIHMS1738032
34217807	Canadian Cardiovascular Society: Clinical Practice Update on Cardiovascular Management of the Pregnant Patient	Windram J, Grewal J, Bottega N, Sermer M, Spears D, Swan L, Siu SC, Silversides C.	Can J Cardiol. 2021 Dec;37(12):1886-1901. doi: 10.1016/j.cjca.2021.06.021. Epub 2021 Jul 1.	Windram J	Can J Cardiol	2021	2021/07/04		
34212413	Dilated ascending aorta in the fetus	Dumitrascu-Biris I, Zidere V, Vigneswaran T, Charakida M, Mathur S, Kametas N, Simpson J.	Prenat Diagn. 2021 Aug;41(9):1127-1133. doi: 10.1002/pd.6007. Epub 2021 Jul 8.	Dumitrascu-Biris I	Prenat Diagn	2021	2021/07/02		
34196822	Atrioventricular Septal Defects: Pathology, Imaging, and Treatment Options	Taatqqa AS, Vettukattil JJ.	Curr Cardiol Rep. 2021 Jul 1;23(8):93. doi: 10.1007/s11886-021-01523-1.	Taatqqa AS	Curr Cardiol Rep	2021	2021/07/01		
34159403	A visual tool inclusive of fetal	Triunfo S, Bonollo M, Gaffuri P.	Arch Gynecol Obstet.	Triunfo S	Arch Gynecol Obstet	2021	2021/06/23		

	ultrasound and autopsy findings to reach a balanced approach to counseling on trisomy 18 in early second trimester	Viviano M, Satta D, Bergmann M.	2021 Nov;304(5): 1115-1125. doi: 10.1007/s00404-021-06130-7.						
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APÊNDICE B - RESULTADOS DE BUSCA NO EMBASE

SEARCH QUERY

('counseling'/exp OR counselling OR 'parent counseling'/exp OR (counseling, AND parente) OR (guidance, AND parente) OR (parent AND guidance)) AND ('congenital heart malformation'/exp OR (congenital AND heart AND anomaly) OR (congenital AND heart AND defect) OR (congenital AND heart AND defects) OR (heart AND anomaly) OR (heart AND congenital AND anomaly) OR (heart AND congenital AND defect) OR (heart AND congenital AND malformation) OR (heart AND defects, AND congenital) OR (heart AND malformation)) AND ('fetus heart'/exp OR (fetal AND heart) OR (foetal AND heart) OR (heart, AND fetus)) AND [embase]/lim AND [humans]/lim

RECORD 1

TITLE

Teen pregnancy in the setting of familial dilated cardiomyopathy: a case report

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BOOK PUBLISHER

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ABSTRACT

Background: Women with pre-existing forms of familial cardiomyopathy are at increased risk for morbidity and mortality due to hemodynamic changes of pregnancy. There is a lack of consensus about the management and care for these patients given the rarity of this condition. This case represents possibly the youngest pregnant familial dilated cardiomyopathy patient to deliver and the youngest patient to be fitted for a wearable cardiac defibrillator in the postpartum period. Case Presentation: A 14-year-old gravida 1 with familial dilated cardiomyopathy presented late for prenatal care at 38 weeks, which precluded typical care plans including baseline and serial echocardiograms, medication management, and routine prenatal care. An echocardiogram showed severely decreased left ventricular systolic function compared to studies from one year prior. Three days later the patient presented in labor and had a spontaneous vaginal delivery complicated by postpartum hemorrhage. Her postpartum course was notable for persistence of decreased cardiac function testing and placement of a wearable cardiac defibrillator for prevention against life threatening arrhythmias. Conclusion: This case report adds to the literature on pregnancy complicated by familial dilated cardiomyopathy and

describes management best practices and considerations during the antepartum, intrapartum, and postpartum periods.

AUTHOR KEYWORDS

Familial Dilated Cardiomyopathy

Prenatal Care

Teen Pregnancy

EMTREE DRUG INDEX TERMS

acetylsalicylic acid (drug therapy, special situation for pharmacovigilance); carboprost (drug therapy, intravenous drug administration, special situation for pharmacovigilance); enalapril (drug therapy, special situation for pharmacovigilance); furosemide (drug therapy, special situation for pharmacovigilance); hemoglobin (endogenous compound); heparin (subcutaneous drug administration); medroxyprogesterone acetate; metoprolol (drug therapy, special situation for pharmacovigilance); misoprostol (drug therapy, special situation for pharmacovigilance); oxytocin (drug therapy, special situation for pharmacovigilance); spironolactone (drug dose, drug therapy, special situation for pharmacovigilance); tranexamic acid (drug therapy, intravenous drug administration, special situation for pharmacovigilance)

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

adolescent pregnancy; congestive cardiomyopathy (diagnosis, drug therapy)

EMTREE MEDICAL INDEX TERMS

abdominal swelling; adolescent; anemia (complication); article; atrial fibrillation; automated external defibrillator; case report; cleft lip; cleft palate; clinical article; contraception; copper intrauterine device; disease severity; drug withdrawal; dyspnea; echocardiography; erythrocyte transfusion; family history; female; fetus heart; gestational age; heart failure; heart left ventricle ejection fraction; heart ventricle septum defect; hemoglobin blood level; hospital discharge; human; labor stage 2; left ventricular end-diastolic diameter; leg swelling; medical history; medication compliance; mitral valve regurgitation; New York Heart Association class; newborn care; patient counseling; patient monitoring; postpartum hemorrhage (complication, drug therapy); prenatal care; puerperium; treatment withdrawal; uterine atony; vaginal delivery

CAS REGISTRY NUMBERS

acetylsalicylic acid (493-53-8, 50-78-2, 53663-74-4, 53664-49-6, 63781-77-1)
carboprost (35700-23-3)
enalapril (75847-73-3)
furosemide (54-31-9)
hemoglobin (9008-02-0)
heparin (37187-54-5, 8057-48-5, 8065-01-8, 9005-48-5, 9041-08-1)
medroxyprogesterone acetate (71-58-9)
metoprolol (37350-58-6)
misoprostol (59122-46-2, 59122-48-4)
oxytocin (50-56-6, 54577-94-5)
spironolactone (52-01-7)
tranexamic acid (1197-18-8, 701-54-2)

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)
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RECORD 2

TITLE

Expanding the phenotype associated with SMARCC2 variants: a fetus with tetralogy of Fallot

AUTHOR NAMES

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ABSTRACT

Background: Coffin-Siris syndrome-8 (CSS8) is a rare autosomal dominant disorder caused by variants in SMARCC2, a core subunit of the chromatin-remodeling complex BRG1-associated factor (BAF). The clinical characteristics of this disorder have not been entirely determined because of the rarity of clinical reports. The BAF complex plays a crucial role in embryogenesis and cardiac development, and pathogenic variants in genes encoding the components of the BAF complex have been associated with congenital heart disease (CHD). However, variants in SMARCC2 have not been reported in patients with CHD. Case presentation: A 28-year-old primigravida was referred at 24 weeks gestation for prenatal echocardiography. The echocardiographic findings were consistent with a prenatal ultrasound diagnosis of tetralogy of Fallot (TOF). After detailed counseling, the couple decided to terminate the pregnancy and undergo genetic testing. A trio (fetus and the parents) whole-exome sequencing (WES) and copy number variation sequencing (CNV-seq) were performed. CNV-seq identified no chromosomal abnormalities. WES analysis revealed a pathogenic, de novo heterozygous frameshift variant in SMARCC2 (NM_003075.5: c.3561del, p.Leu1188fs). The genetic diagnosis of CSS8 was considered given the identification of the SMARCC2 pathogenic variant. Conclusions: We report the first prenatal case with the SMARCC2 variant. The presence of CHD further broadens the phenotypic spectrum of SMARCC2-related disease.

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

Fallot tetralogy; phenotype

EMTREE MEDICAL INDEX TERMS

adult; article; case report; chromosome aberration; clinical article; congenital heart disease; copy number variation; counseling; diagnosis; echocardiography; female; fetus; fetus echography; frameshift mutation; genetic screening; heterozygosity; human; pregnancy; primigravida; whole exome sequencing

LANGUAGE OF ARTICLE

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LANGUAGE OF SUMMARY

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RECORD 3**TITLE**

Novel large deletion involving EVC and EVC2 in Ellis–van Creveld syndrome

AUTHOR NAMES

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BOOK PUBLISHER

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ABSTRACT

Ellis–van Creveld syndrome is an autosomal recessive skeletal dysplasia that is characterized by thoracic hypoplasia, polydactyly, oral abnormalities, and congenital heart disease. It is caused by pathogenic variants in the EVC or EVC2 genes. We report a case of a newborn with a compound heterozygous variant comprising NM_147127.5: c.1991dup:[p.Lys665Glufs*10] in the EVC2 gene and a novel large deletion involving exon 1 in EVC and exons 1–7 in EVC2.

EMTREE DRUG INDEX TERMS (MAJOR FOCUS)

ellis van crefeld syndrome ciliary complex subunit 1 protein (endogenous compound); ellis van crefeld syndrome ciliary complex subunit 2 protein (endogenous compound); membrane protein (endogenous compound)

EMTREE DRUG INDEX TERMS

genomic DNA (endogenous compound); nitric oxide; oxytocin; sedative agent (special situation for pharmacovigilance); unclassified drug

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

Ellis van Creveld syndrome (diagnosis, etiology); gene deletion; genetic association; newborn disease (diagnosis, etiology)

EMTREE MEDICAL INDEX TERMS

Apgar score; article; artificial ventilation; atrioventricular septal defect; bilateral polydactyly; birth length; body height; body weight; c4orf6 gene; cardiac imaging; case report; clavicle fracture; clinical article; developmental delay; disease severity; DNA sequencing; drug withdrawal; ductus arteriosus; dystocia; enteric feeding; exon; fetus echography; frameshift mutation; gene frequency; genetic counseling; genetic screening; genetic variability; gestational age; growth disorder; head circumference; heart right left shunt; heterozygote; high flow nasal cannula therapy; high throughput sequencer; home oxygen therapy; hospital discharge; human; karyotype 46,XY; labor induction; lung hypoplasia; male; multiplex ligation dependent probe amplification; nail dysplasia; neonatal intensive care unit; neonatal resuscitation; newborn; newborn hypoxia (diagnosis, therapy); NextSeq2000; pathogenesis; persistent pulmonary hypertension (therapy); phenotype; polydactyly; primipara; quantitative analysis; resuscitation; Sanger sequencing; single nucleotide polymorphism; stk32b gene; thorax radiography; tooth malformation; tricuspid valve regurgitation

DEVICE TRADE NAMES

NextSeq2000, United States

DEVICE MANUFACTURERS

(United States)Illumina

CAS REGISTRY NUMBERS

nitric oxide (10102-43-9)

oxytocin (50-56-6, 54577-94-5)

EMBASE CLASSIFICATIONS

Human Genetics (22)

Clinical and Experimental Biochemistry (29)

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RECORD 4

TITLE

Infantile neurodevelopmental outcome after fetoscopic laser photocoagulation for twin-to-twin transfusion syndrome: the first prospective experience from Iran

AUTHOR NAMES

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ABSTRACT

Objective: We sought to evaluate the neurodevelopmental outcomes at 12 months of age among infants with twin-to-twin transfusion syndrome (TTTS) undergoing fetoscopic laser photocoagulation (FLP). Materials and methods: In this prospective longitudinal study, neurodevelopmental assessment was performed among the infants at the corrected age of 12 months, who were diagnosed with TTTS and treated by FLP. The Ages and Stages Questionnaire (ASQ) was filled out by parents. In the next step in infants with abnormal ASQ, motor and cognitive developments were evaluated by Bayley's infant and toddler development scoring system (Bayley 3-Third edition). Results: In 39 FLP procedures the rate of live birth of at least one twin was 73.8%. Four neonatal deaths were recorded, three of which were due to prematurity and one was due to heart anomaly. The ASQ was normal in 89.7% (35/39) of the infants (group I), 5.1% (2/39) had minor neurodevelopmental impairment (NDI) (group II), and 5.1% (2/39) had major NDI (group III). The 4 infants with abnormal ASQ had Bayley examination which showed two with mild to moderate cerebral palsy and two had delayed verbal skills and autistic spectrum disorder. No significant difference was noted between survivors with and without NDI with respect to donor or recipient status, birth weight, gestational age at birth, Quintero stage of TTTS. In addition, the relationship between gestational age at the time of undergoing FLC and NDI was not significant. Conclusion: In our population, minor and major neurodevelopmental impairment were seen in 10.2% of the infants. This information is useful for counseling our couples in this population prior the procedure.

AUTHOR KEYWORDS

Fetoscopic laser photocoagulation

Monochorionic

Neurodevelopment

Twin pregnancy

Twin twin transfusion syndrome

EMTREE DRUG INDEX TERMS

ceftriaxone (intravenous drug administration); hydroxyprogesterone caproate (intramuscular drug administration); indometacin (rectal drug administration); nifedipine (oral drug administration)

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

child development; Iran; laser coagulation; nervous system development; twin twin transfusion syndrome (surgery)

EMTREE MEDICAL INDEX TERMS

age; Ages and Stages Questionnaire; article; autism (complication); Bayley Scales of Infant Development; birth weight; cerebral palsy (complication); clinical article; cognitive development; congenital heart malformation; gestational age; human; infant; language delay (complication); live birth; longitudinal study; mental development assessment; mental disease (complication); motor development; newborn death; parent counseling; prematurity; prospective study; survivor

CAS REGISTRY NUMBERS

ceftriaxone (73384-59-5, 74578-69-1)
hydroxyprogesterone caproate (630-56-8)
indometacin (53-86-1, 74252-25-8, 7681-54-1)
nifedipine (21829-25-4)

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)
Drug Literature Index (37)
Pediatrics and Pediatric Surgery (7)
Neurology and Neurosurgery (8)

LANGUAGE OF ARTICLE

English

LANGUAGE OF SUMMARY

English

MEDLINE PMID

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DOI

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EMBASE LINK

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RECORD 5**TITLE**

Prenatal diagnosis and genetic counseling of a paternally inherited chromosome 15q11.2 microdeletion in a Chinese family

AUTHOR NAMES

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15

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Article

ISSN

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BOOK PUBLISHER

BioMed Central Ltd

ABSTRACT

Background: Proximal region of chromosome 15 long arm is rich in duplicons that, define five breakpoints (BP) for 15q rearrangements. 15q11.2 microdeletion has been previously associated with developmental delay, mental retardation, epilepsy, autism, schizophrenia and congenital heart defects. The literature on this microdeletion is extensive and confusing, which is a challenge for genetic counselling. Case presentation: We have performed prenatal diagnosis and genetic counseling of a paternally inherited 15q11.2 microdeletion. In this family, father with normal phenotype and fetus with abnormal phenotype have the same microdeletion. Conclusion: Chromosomal microdeletions and microduplications are difficult to detect

by conventional cytogenetics, combination of prenatal ultrasound, karyotype analysis, CMA and genetic counseling is helpful for the prenatal diagnosis of chromosomal microdeletions/microduplications.

AUTHOR KEYWORDS

Chromosomal microarray analysis (CMA)

Chromosomal microdeletions/microduplications

Prenatal diagnosis

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

Chinese; chromosome 15q; genetic counseling; microarray analysis; paternal inheritance; prenatal diagnosis

EMTREE MEDICAL INDEX TERMS

article; case report; clinical article; cytogenetics; deletion mutant; diagnosis; father; fetus; fetus echography; gene deletion; human; human experiment; karyotyping; male; phenotype

LANGUAGE OF ARTICLE

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LANGUAGE OF SUMMARY

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RECORD 6

TITLE

Maternal Health: The Heart of the Matter

AUTHOR NAMES

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AI/IP ENTRY DATE

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BOOK PUBLISHER

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EMTREE DRUG INDEX TERMS

amino terminal pro brain natriuretic peptide (endogenous compound)

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

maternal welfare

EMTREE MEDICAL INDEX TERMS

abortion; adverse outcome; African American; all cause mortality; anemia; angiography; aortic dissection; atherosclerosis; attitude to health; birth rate; blood pressure variability; blood volume; California; Canada; cardiologist; cardiology; cardiovascular disease; cardiovascular mortality; Caucasian; cerebrovascular accident; clinical decision making; comparative study; congenital heart disease; coronary artery dissection; coronavirus disease 2019 (epidemiology); demographics; disease classification; disease course; disease predisposition; disease severity; eclampsia; editorial; endothelial dysfunction; essential hypertension; fetus mortality; France; health care access; health care personnel; health care policy; health disparity; health equity; health insurance; health statistics; heart atrium arrhythmia; heart failure; heart infarction; heart left ventricle hypertrophy; heart output; heart single ventricle; heart ventricle arrhythmia; heart ventricle remodeling; human; inferior cava vein obstruction; inflammation; intersectoral collaboration; legal aspect; live birth; maternal age; maternal care; maternal diabetes mellitus; maternal hypertension; maternal morbidity; maternal mortality; maternal obesity; maternal stress; maternity ward; medical education; medical society; mortality risk; multidisciplinary team; obstetric delivery; obstetrics; pandemic; patient attitude; patient counseling; patient referral; patient triage; peripartum cardiomyopathy; placenta disorder; practice guideline; prediction; preeclampsia; pregnancy complication; pregnancy termination; prenatal care; prevalence; publication; puerperium; pulmonary hypertension; reproductive health; reproductive rights; risk assessment; social determinants of health; systemic vascular resistance; third trimester pregnancy; United States; valvular heart disease (congenital disorder); venous thromboembolism; women's health

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

Chest Diseases, Thoracic Surgery and Tuberculosis (15)
Public Health, Social Medicine and Epidemiology (17)
Cardiovascular Diseases and Cardiovascular Surgery (18)
Microbiology: Bacteriology, Mycology, Parasitology and Virology (4)
Pediatrics and Pediatric Surgery (7)

LANGUAGE OF ARTICLE

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MEDLINE PMID

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RECORD 7

TITLE

Atrial Appendage Anastomosis in Hypoplastic Left Heart Syndrome With Restrictive Atrial Septum: A Novel Approach

AUTHOR NAMES

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Article

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BOOK PUBLISHER

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ABSTRACT

Intact atrial septum (IAS), occurring in ~10% of patients with hypoplastic left heart syndrome (HLHS), conveys significant neonatal morbidity and mortality. Perinatal interventions have been described, but outcomes remain poor. We present a fetus with HLHS with IAS who underwent immediate novel postnatal atrial appendage anastomosis, thus achieving rapid left atrial decompression. (Level of Difficulty: Advanced.)

AUTHOR KEYWORDS

atrial appendage anastomosis
hypoplastic left heart syndrome
intact atrial septum

EMTREE DRUG INDEX TERMS

diuretic agent (drug therapy)

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

anastomosis; heart atrium appendage; heart atrium septum defect (diagnosis, surgery); hypoplastic left heart syndrome (diagnosis, surgery); restrictive atrial septum (diagnosis, surgery)

EMTREE MEDICAL INDEX TERMS

adult; aortic stenosis (diagnosis, surgery); aortic valve repair; article; cardiopulmonary bypass; case report; cavopulmonary connection; cesarean section; clamp; clinical article; counseling; disease exacerbation; disease severity; echocardiography; female; fetus; fetus echography; heart catheterization; human; labor; labor induction; lymphangiectasis (drug therapy); mitral valve regurgitation (surgery); multidisciplinary team; obstetric delivery; operating room; oxygen saturation; perinatal period; pregnancy termination; premature labor; spontaneous abortion; sternotomy; tricuspid valve repair

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)
Radiology (14)
Cardiovascular Diseases and Cardiovascular Surgery (18)
Drug Literature Index (37)

LANGUAGE OF ARTICLE

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LANGUAGE OF SUMMARY

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RECORD 8

TITLE

Prenatal diagnosis and postnatal outcomes of right aortic arch anomalies

AUTHOR NAMES

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Archives of Gynecology and Obstetrics

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DATE OF PUBLICATION

1 Sep 2022

PUBLICATION TYPE

Article

ISSN

1432-0711 (electronic)

0932-0067

BOOK PUBLISHER

Springer Science and Business Media Deutschland GmbH

ABSTRACT

Purpose: To give a report on the experience of our tertiary perinatology clinic on the pre- and postnatal management of the right aortic arch (RAA) by evaluating the patients as isolated and non-isolated RAA. **Materials and methods:** Patients referred to our perinatology clinic for fetal echocardiography were evaluated retrospectively. They were assessed in two groups: isolated RAA and non-isolated RAA. The isolated RAA group consisted of patients without any additional cardiac or extracardiac anomalies. According to our routine practice, all patients received detailed prenatal ultrasonography following fetal echocardiography and genetic counseling. **Results:** A total of 60 patients were evaluated. 38 patients (63.3%) presented with additional cardiac anomalies. 21.7% had extracardiac anomalies, including 16.7% who also had cardiac anomalies. In 2 patients (3.7%) 22q11.2 microdeletion, in 2 patients (3.7%) trisomy 21, in 1 patient (1.9%) trisomy 13 and in 1 patient (1.9%) 20p12.1p11.23 (a deletion of 2880 kbp) were reported. The most common cardiac anomaly associated with RAA was Tetralogy of Fallot (25%). Fetal growth restriction was reported in 8.3% of the cases. 18 patients had isolated RAA. 16 out of the 18 patients had normal genetic analysis. 2 of them (11.11%) presented with a 22q11.2 microdeletion. **Conclusion:** A single-center experience on the diagnosis and management of RAA has been reported in this study. The results indicate that a prenatal cardiac evaluation in 3VV is of utmost importance in all pregnancies to detect RAA and refer these patients to the appropriate perinatology clinics for further evaluation and care.

AUTHOR KEYWORDS

22q11.2 microdeletion

Congenital heart disease

Fetal echocardiography

Right aortic arch

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

aortic arch anomaly; echocardiography; Fallot tetralogy; fetus echography

EMTREE MEDICAL INDEX TERMS

adult; article; clinical evaluation; congenital heart disease; diagnosis; female; gene deletion; genetic analysis; genetic counseling; human; intrauterine growth retardation; major clinical study; perinatology; pregnancy; retrospective study; trisomy 13; trisomy 21

LANGUAGE OF ARTICLE

English

LANGUAGE OF SUMMARY

English

MEDLINE PMID

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RECORD 9**TITLE**

Maternal and Fetal Outcomes in Women with Congenital Heart Disease

AUTHOR NAMES

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DATE OF PUBLICATION

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PUBLICATION TYPE

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1053-0770

BOOK PUBLISHER

W.B. Saunders

EMTREE DRUG INDEX TERMS

warfarin

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

congenital heart disease; fetus outcome; pregnancy outcome

EMTREE MEDICAL INDEX TERMS

algorithm; anesthesiologist; anticoagulation; cardiologist; editorial; Fallot tetralogy; female; Fontan procedure; functional status; heart atrium arrhythmia; heart disease; heart function; heart output; human; junctional ectopic tachycardia; maternal care; patient counseling; postpartum hemorrhage; pregnancy complication; pregnant woman; premature labor; prenatal care; third trimester pregnancy

CAS REGISTRY NUMBERS

warfarin (129-06-6, 2610-86-8, 3324-63-8, 5543-58-8, 81-81-2)

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

Cardiovascular Diseases and Cardiovascular Surgery (18)

Drug Literature Index (37)

Pediatrics and Pediatric Surgery (7)

LANGUAGE OF ARTICLE

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MEDLINE PMID

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EMBASE LINK

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TITLE

Antenatal findings and early postnatal outcomes in pregnancies with trisomy 21: a 10 year retrospective review at a tertiary centre

AUTHOR NAMES

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PUBLICATION TYPE

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0197-3851

BOOK PUBLISHER

John Wiley and Sons Ltd

ABSTRACT

Objective: To examine the antenatal imaging features, intrapartum findings and early postpartum course of pregnancies with trisomy 21 (T21) at a tertiary hospital in the United Kingdom. **Methods:** Women with pregnancies diagnosed with T21 on antenatal or post-mortem/postnatal karyotyping, from February 2010–2020. Outcome measures included antenatal imaging findings, fetal growth restriction (FGR), birthweight, mode of delivery and early neonatal outcomes. **Results:** 76 women were included. There were six intrauterine deaths and 70 livebirths. Thirty-eight (50%) had an antenatal diagnosis and twenty-five (33%) had a suspected diagnosis but declined further testing. The diagnosis was unanticipated in 13 (17%). Cardiac anomalies (35.5%) were the most common antenatal anomaly. Doppler abnormalities were apparent in 48/73 (68%). Eighteen (25.7%) had antenatal FGR. The majority were delivered by Caesarean section, and 21.4% of babies weighed <third percentile at delivery. Fifty-eight (82%) were admitted to the neonatal unit. Forty-three (61%) required respiratory support and fifty-five (78%) needed naso-gastric feeding or were nil by mouth. Mean PAPP-A values were significantly lower in cases with abnormal Dopplers, FGR, congenital anomalies and birthweight <10th percentile. **Conclusions:** T21 fetuses have high rates of placental insufficiency, FGR and Doppler abnormalities. Postnatally, most require respiratory and feeding support. Antenatal counselling should reflect these risks.

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

outcome assessment; trisomy 21 (diagnosis)

EMTREE MEDICAL INDEX TERMS

article; assisted ventilation; birth weight; cesarean section; clinical feature; female; fetus; fetus death; fetus growth; fetus outcome; heart disease; human; intrauterine growth retardation; karyotyping; major clinical study; obstetric delivery; perinatal period; pregnancy; prenatal diagnosis; prenatal period; puerperium; retrospective study; ultrasound; United Kingdom

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

Radiology (14)

LANGUAGE OF ARTICLE

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LANGUAGE OF SUMMARY

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MEDLINE PMID

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RECORD 11**TITLE**

Assisted reproduction in patients with cardiac disease: A retrospective review

AUTHOR NAMES

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SOURCE

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European Journal of Obstetrics and Gynecology and Reproductive Biology

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0301-2115

BOOK PUBLISHER

Elsevier Ireland Ltd

ABSTRACT

Objective: To assess risks of assisted reproduction in patients with cardiac disease. Study design: Retrospective case note review of patients with cardiac disease undergoing ART over a 10 year period in the obstetric cardiac services of three UK tertiary centres. Assessment of maternal, obstetric and fetal complications during ART and resultant pregnancies. Results: 34 patients with cardiac disease underwent 51 cycles of assisted reproduction. 24 patients (71%) received pre-pregnancy counselling. Mean age at the start of an assisted reproduction cycle was 32 years. Modified WHO (mWHO) risk category for the 34 patients was mWHO I, n = 3; mWHO II, n = 13; mWHO II- III, n = 10; mWHO III, n = 7; mWHO IV, n = 1. The 51 assisted reproduction cycles resulted in 31 pregnancies in 29 patients, and 31 live births, including two sets of twins. Live birth rate per cycle was 60.8%. Twin pregnancy rate per cycle was 5.8%. Four patients experienced complications during assisted reproduction treatment (7.8% per cycle); one major intra-abdominal haemorrhage following egg collection in a patient with a mechanical aortic valve, one endocarditis, one mild ovarian hyperstimulation syndrome and one vagal syncope during egg collection. Four other patients experienced cardiac complications during resultant pregnancies (12.9%). 43% of mWHO class III patients experienced cardiac, obstetric or neonatal complications. Five babies were delivered pre-term (<37/40). Conclusions: This small study demonstrates that assisted reproduction carries increased risks of complications in patients with cardiac disease, but can be undertaken without major complication in the majority, as long as appropriate adjustments to treatment pathways are made, and they are managed through a multi-disciplinary team.

AUTHOR KEYWORDS

Cardiac disease

Medically assisted reproduction

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

heart disease; infertility therapy; reproduction; risk assessment

EMTREE MEDICAL INDEX TERMS

abdominal bleeding; adult; article; atrioventricular nodal reentry tachycardia; bicuspid aortic valve; clinical article; congenital heart malformation (surgery); controlled study; dyspnea; endocarditis; estrus cycle; faintness; Fallot tetralogy; female; heart atrium septum defect; heart failure; heart ventricle tachycardia; hemoperitoneum; human; live birth; maternal hypertension; mitral valve regurgitation; multiple pregnancy; ovary hyperstimulation; preeclampsia; pregnancy complication; pregnancy outcome; pulmonary valve stenosis; restrictive cardiomyopathy; retrospective study; small for date infant; Turner syndrome; twin pregnancy; uterus bicornis

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

Cardiovascular Diseases and Cardiovascular Surgery (18)

LANGUAGE OF ARTICLE

English

LANGUAGE OF SUMMARY

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MEDLINE PMID

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RECORD 12

TITLE

Case report: Fetal cervical immature teratoma and copy number variations

AUTHOR NAMES

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Frontiers Media S.A.

ABSTRACT

Fetal cervical teratoma is a rare congenital neck tumor. Here, we report a case of a fetus with an anterior solid neck tumor that was confirmed to have an immature teratoma by histology. A duplication was found at chromosome 14q24.1-q24.3 of the fetus in chromosome microarray (CMA) and whole exome sequencing (WES), which was a copy number variation (CNV) and a probably new-onset. Ultrasound coupled with magnetic resonance imaging (MRI) can be considered to be a relatively reliable diagnostic tool, whereas ex-utero intrapartum therapy or resection of the tumor mass on placental support may improve the chances of the newborn's survival. Strangely, the same duplication occurred on her next fetus that was found with complex congenital heart malformations. CNV at chromosome 14q24.1-q24.3 needs to be paid more attention.

AUTHOR KEYWORDS

chromosome microarray

copy number variations

mutation

teratoma

whole exome sequencing

EMTREE DRUG INDEX TERMS

chorionic gonadotropin (endogenous compound)

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

cervical teratoma (diagnosis); copy number variation

EMTREE MEDICAL INDEX TERMS

adult; amniocentesis; article; automated tissue processor; autopsy; case report; chromosome 14q; chromosome analysis; clinical article; comparative genomic hybridization; congenital heart malformation; cytogenetics; DNA microarray;

echography; eye fundus; female; fetus; fetus echography; gene mutation; genetic analysis; genetic counseling; genetic screening; high throughput sequencer; high throughput sequencing; histology; histopathology; human; human tissue; intensive care unit; karyotype; lung dysplasia; microarray analysis; microcephaly; microtome; muscle hypotonia; nuclear magnetic resonance imaging; nucleic acid isolation kit; placenta; pleura effusion; pregnancy diabetes mellitus; premature labor; prenatal diagnosis; trisomy; ultrasound; whole exome sequencing

DEVICE MANUFACTURERS

(Canada)Illumina

(Germany)Leica

Magen

(Germany)Micromed

CAS REGISTRY NUMBERS

chorionic gonadotropin (9002-61-3)

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

Human Genetics (22)

Biophysics, Bioengineering and Medical Instrumentation (27)

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LANGUAGE OF ARTICLE

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RECORD 13

TITLE

Perinatal Outcomes of Fetuses and Infants Diagnosed with Trisomy 13 or Trisomy 18

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BOOK PUBLISHER

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ABSTRACT

Objectives: To identify factors associated with prenatal, perinatal, and postnatal outcomes, and determine medical care use for fetuses and infants with trisomy 13 (T13) and trisomy 18 (T18). **Study design:** This population-based retrospective cohort study included all prenatal and postnatal diagnoses of T13 or T18 in the greater Cincinnati area from January 1, 2012, to December 31, 2018. Overall survival, survival to hospital discharge, medical management, and maternal, fetal, and neonatal characteristics are analyzed. **Results:** There were 124 pregnancies (125 fetuses) that were identified, which resulted in 72 liveborn infants. Male fetal sex and hydrops were associated with a higher rate of spontaneous loss. The median length of survival was 7 and 29 days, for infants with T13 and T18, respectively. Of the 27 infants alive at 1 month of age, 13 (48%) were alive at 1 year of age. Only trisomy type (T13), goals of care (comfort care), and extremely low birthweight were associated with a shorter length of survival. A high degree of variability existed in the use of medical services, with 28% of infants undergoing at least 1 surgical procedure and some children requiring repeated (≤ 29) or prolonged (>1 year) hospitalizations. **Conclusions:** Although many infants with T13 or T18 did not survive past the first

week of life, nearly 20% lived for more than 1 year with varying degrees of medical support. The length of survival for an infant cannot be easily predicted, and surviving infants have high health care use throughout their lifespans.

AUTHOR KEYWORDS

congenital abnormalities

neonatal intensive care

prenatal diagnosis

trisomy 18

trisomy13

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

prenatal diagnosis; trisomy 13 (congenital disorder, diagnosis); trisomy 18 (congenital disorder, diagnosis)

EMTREE MEDICAL INDEX TERMS

adult; article; artificial ventilation; assisted ventilation; child hospitalization; cohort analysis; disease management; fetus; fetus hydrops; genetic counseling; genetic screening; gestational age; heart atrium septum defect (congenital disorder, diagnosis); heart ventricle septum defect (congenital disorder, diagnosis); hospital discharge; human; infant; live birth; low birth weight; major clinical study; male; maternal age; median survival time; medical care; medical record review; medical service; monochorionic diamniotic twins; overall survival; patent ductus arteriosus (congenital disorder, diagnosis); perinatal period; postnatal care; pregnancy; pregnancy outcome; prenatal period; retrospective study; spontaneous abortion; stomach tube

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

Anesthesiology (24)

Pediatrics and Pediatric Surgery (7)

LANGUAGE OF ARTICLE

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RECORD 14**TITLE**

Prenatal diagnosis of Miller-Dieker syndrome/PAFAH1B1-related lissencephaly: Ultrasonography and genetically investigative results

AUTHOR NAMES

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0301-2115

BOOK PUBLISHER

Elsevier Ireland Ltd

ABSTRACT

Objective: To present the experience on prenatal diagnosis of Miller-Dieker syndrome (MDS)/PAFAH1B1-related lissencephaly to further determine fetal phenotypes of this syndrome. Study design: This was a retrospective study of ten pregnancies with fetal MDS/PAFAH1B1-related lissencephaly identified by chromosomal microarray (CMA)/exome sequencing (ES). Clinical and laboratory data were collected and reviewed for these cases, including maternal demographics, prenatal sonographic findings, CMA or ES results and pregnancy outcomes. Results: Two cases were diagnosed in the first trimester because of an increased nuchal translucency. The remaining eight cases were identified at late gestation, including four in the second trimester because of fetal cardiac anomalies or ventriculomegaly, and four in the third trimester because of ventriculomegaly. CMA revealed 17p13.3

deletions in nine cases, and ES detected a de novo PFAH1B1 missense mutation in one case. Conclusion: The prenatal presentation of MDS/PFAH1B1-related lissencephaly depended on the gestational age when the diagnosis was made. Mild ventriculomegaly was the most common prenatal sonographic sign identified in cases of MDS/PFAH1B1-related lissencephaly. It is important that fetal MRI and invasive testing with CMA should be considered in fetuses with apparently 'isolated' mild ventriculomegaly.

AUTHOR KEYWORDS

Lissencephaly
Miller-Dieker syndrome
PFAH1B1 gene
Prenatal diagnosis
Ventriculomegaly

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

agyria (diagnosis, etiology); gene; Miller Dieker syndrome (diagnosis, etiology); PFAH1B1 gene; prenatal diagnosis

EMTREE MEDICAL INDEX TERMS

adult; amniocentesis; article; brain ventricle dilatation (diagnosis); chromosome analysis; chromosome deletion; chromosome deletion 17p13.3; clinical article; congenital heart malformation (diagnosis); demographics; female; fetus; fetus disease (diagnosis); fetus echography; first trimester pregnancy; fluorescence quantitative polymerase chain reaction; GE E10; GE E8; genetic counseling; gestational age; human; hydramnios (diagnosis); intrauterine growth retardation (diagnosis); karyotyping; macrogyria; maternal age; missense mutation; MRI scanner; nuchal translucency measurement; nuclear magnetic resonance imaging; phenotype; pregnancy outcome; retrospective study; second trimester pregnancy; third trimester pregnancy; whole exome sequencing

DEVICE TRADE NAMES

GE E10, United States
GE E8, United States

DEVICE MANUFACTURERS

(United States)General Electric

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

Radiology (14)

Human Genetics (22)

General Pathology and Pathological Anatomy (5)

Pediatrics and Pediatric Surgery (7)

Neurology and Neurosurgery (8)

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RECORD 15

TITLE

Persistent Left Superior Vena Cava Significance in Prenatal Diagnosis—Case Series

AUTHOR NAMES

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BOOK PUBLISHER

MDPI

ABSTRACT

The persistent left superior vena cava (PLSVC) is a congenital heart anomaly reported in 0.3–0.5% of the general population and can be associated with congenital heart diseases in up to 8% of cases. Prenatal identification of PLSVC is important to prompt an extended cardiac and extracardiac fetal examination. We retrospectively reevaluated anomaly scans performed in our unit in a 2-year interval according to the national guidelines to evaluate the incidence of PLSVC and its association with prenatal morbidity. In our population, the incidence of PLSVC was 0.31%, and we found a low association with cardiac and extracardiac anomalies. The standard sections (three-vessel and trachea view, four-chamber view and outflow tract's view) are insufficient to exclude cardiac anomalies whenever PLSVC is found. In our case series, only one newborn required postnatal surgery for total pulmonary vein anomaly, and at 2 years of life all babies had a normal evolution. Prenatal diagnosis of PLSVC can raise counseling issues; therefore, awareness of its good outcome when isolated and need for an extended examination to rule out other anomalies is very important.

AUTHOR KEYWORDS

congenital heart disease
congenital venous anomaly
persistent left superior vena cava
prenatal diagnosis

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

case study; congenital heart disease; persistent left superior vena cava; prenatal diagnosis

EMTREE MEDICAL INDEX TERMS

article; awareness; congenital heart malformation; counseling; diagnosis; human; incidence; newborn; outcome assessment; pulmonary vein malformation; surgery; trachea

LANGUAGE OF ARTICLE

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RECORD 16**TITLE**

Prenatal diagnosis and molecular cytogenetic characterization of inherited chromosome 2q11.1q11.2 microduplication with fetal intrauterine growth retardation

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EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

chromosome 2q; chromosome duplication; intrauterine growth retardation; prenatal diagnosis

EMTREE MEDICAL INDEX TERMS

adult; amniocentesis; article; case report; cell culture; chromosome deletion; clinical article; cytogenetic analysis; face dysmorphia; fetus echography; genetic counseling; heart ventricle septum defect; human; human cell; human tissue; intellectual impairment; intravenous catheter; karyotyping; microarray analysis; microcephaly; partial trisomy; pregnancy termination

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

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Human Genetics (22)

Pediatrics and Pediatric Surgery (7)

LANGUAGE OF ARTICLE

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RECORD 17**TITLE**

Prenatal Diagnosis and Management of Single-Ventricle Heart Disease

AUTHOR NAMES

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Focus Issue: Single Ventricle Heart Disease Across the Lifespan, Book Series Title:

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Canadian Journal of Cardiology

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Review

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BOOK PUBLISHER

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ABSTRACT

In the current era, most single-ventricle heart disease (SVHD) is diagnosed prenatally by means of fetal echocardiography. Disparities exist, however, by socioeconomic status and remote location, which require further attention. Prenatal diagnosis affords the opportunity to counsel expectant parents regarding the life-long

course of children with SVHD, including the stages of single-ventricle palliation and challenges of the Fontan circulation; to discuss pregnancy management options; and to optimise delivery planning and perinatal care. Prognosis may be refined by specific features on the fetal echocardiogram, such as ventricular morphology, total anomalous pulmonary venous return, and atrioventricular valve regurgitation. Expectant mothers should be referred for evaluation of extracardiac anomalies and/or a genetic syndrome, which also significantly affect outcome. Fetuses with SVHD should be cared for by a multidisciplinary team and ideally delivered at term at or near a cardiac surgical center. Serial echocardiograms refine the anticipated postnatal physiology to optimise transitional care, including the need for prostaglandin or urgent atrial septal intervention in fetuses with hypoplastic left heart syndrome. In selected patients, there may be a role for fetal cardiac intervention to improve mortality or achieve a biventricular circulation after birth. Together, these strategies enhance the preoperative status of the neonate. Recent advances in fetal cardiovascular magnetic resonance imaging have focused on studying the relationships between cardiovascular physiology and fetal growth and development. These novel techniques allow for the exploration of the physiologic effects of SVHD on the brain and open avenues for the investigation of neuroprotective therapies.

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

heart single ventricle (diagnosis, surgery); prenatal diagnosis

EMTREE MEDICAL INDEX TERMS

cardiovascular magnetic resonance; child; clinical outcome; counseling; diagnostic imaging; echocardiography; fetus; fetus echography; genetic disorder; human; perinatal care; pregnancy; pulmonary valvuloplasty; review; septoplasty

EMBASE CLASSIFICATIONS

Radiology (14)

Cardiovascular Diseases and Cardiovascular Surgery (18)

Pediatrics and Pediatric Surgery (7)

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RECORD 18

TITLE

Reproductive Issues in Patients With the Fontan Operation

AUTHOR NAMES

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ABSTRACT

Patients with the Fontan operation have a unique circulation, with a limited ability to increase cardiac output, and high central venous pressure. They may have diastolic and/or systolic ventricular dysfunction, arrhythmias, thromboembolic complications, or multiorgan dysfunction. All of these factors contribute to reproductive issues, including menstrual irregularities, infertility, recurrent miscarriage, and complications during pregnancy. Although atrial arrhythmias are the most common cardiac

complications during pregnancy, patients can develop heart failure and thromboembolic events. Obstetric bleeding, including postpartum hemorrhage, is common. In addition to maternal complications, adverse fetal and neonatal events, such as prematurity and low birthweight, are very common. Counselling about these reproductive issues should begin early. For those who become pregnant, care should be provided by a multidisciplinary cardio-obstetric team familiar with the specific issues and needs of the Fontan population. In this review, we discuss infertility, contraception, and pregnancy in patients with the Fontan operation.

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

Fontan procedure; gynecologic disease

EMTREE MEDICAL INDEX TERMS

cardiovascular risk; contraception; female infertility; fetus risk; heart atrium arrhythmia; heart failure; heart single ventricle (surgery); hemodynamics; high risk pregnancy; human; infertility therapy; low birth weight; menstrual irregularity; multidisciplinary team; patient counseling; postpartum hemorrhage; pregnancy complication; prematurity; prepregnancy care; review; spontaneous abortion; thromboembolism

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

Cardiovascular Diseases and Cardiovascular Surgery (18)

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RECORD 19

TITLE

Fetal cystic hygroma: Prenatal diagnosis and postnatal outcome

AUTHOR NAMES

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AUTHOR ADDRESSES

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CONFERENCE DATE

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BOOK PUBLISHER

Malaysian Medical Association

ABSTRACT

Introduction: Cystic Hygroma occurs due to lymphatic malformation in different parts of the fetus. We present a case of late detection of huge multiseptated cystic hygroma with hydrops fetalis at 28 weeks gestation and intrapartum outcome. Late detection of cystic hygroma may give rise to a dilemma in decision-making due to considerations of cephalopelvic disproportion and unnecessary caesarean sections. Case Description: We report a case of a 32-year-old, Gravida 5, Para 2 at 28 weeks who presented in preterm labour. At 18 weeks gestation, the fetus was suspected to have a fetal neck mass. However, she defaulted follow-ups and had no further assessment until she presented in labour. Ultrasonography revealed a fetus in breech presentation, with generalized oedema and hydrops fetalis features. There was a cystic hygroma measuring 13.2 x 10.2 cm. There was no fetal heart pulsation and an intrauterine demise was diagnosed. Induction of labour was performed with an anticipation of head entrapment in the second stage due to the presence of cystic hygroma. The head entrapment that eventually occurred was resolved with Mauriceau manoeuvre technique, and a stillborn baby boy weighing 1900 gram was delivered. Discussion: An alternative method to aid delivery of the head in the presence of a large cystic hygroma is by an ultrasound-guided decompression of the cystic mass using a spinal needle, should the usual manoeuvre to deliver the aftercoming head fails. Early detection of cystic hygroma and hydrops fetalis during

the prenatal period may allow for proper intrapartum management and parental counselling.

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

fetal cystic hygroma; prenatal diagnosis

EMTREE MEDICAL INDEX TERMS

adult; anticipation; breech presentation; case report; cephalopelvic disproportion; cesarean section; child; clinical article; conference abstract; counseling; decision making; decompression; diagnosis; echography; female; fetus; fetus heart; fetus hydrops; generalized edema; human; male; neck tumor; pregnancy; premature labor; prenatal period; spinal needle; stillbirth

LANGUAGE OF ARTICLE

English

LANGUAGE OF SUMMARY

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RECORD 20

TITLE

Omphalocele and umbilical cord cyst: A case report and review of literature

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Malaysian Medical Association

ABSTRACT

Introduction: Omphalocele is a midline defect with evisceration of abdominal contents covered by a protective sac. The lack of knowledge about the prenatal diagnosis of omphalocele and umbilical cord cyst may lead to missed diagnosis by practitioners. For proper intrapartum management and parental counselling,

associated anomalies should be confidently ruled out during the prenatal period. Case Description: We report a case of a 41-year-old, Gravida 6, Para 2 + 2 at 24 weeks with prenatal ultrasonography findings of huge umbilical cord cyst measuring 6.5 x 6.3 cm, an omphalocele with liver content with cord insertion at the omphalocele measuring 4.5 x 4.5 cm, a choroid plexus cyst and a small thorax with hypoplastic left ventricle and bilateral lungs. Given a guarded fetal prognosis, the pregnancy was terminated after careful discussion with the parents. A 500-gram male fetus was delivered and expired immediately. The fetus has a substantial anterior abdominal wall defect with liver content within the omphalocele, a uniloculated umbilical cord cyst containing Wharton Jelly and straw-coloured fluid, bilateral clenched fists and micrognathia. A clinical diagnosis of Edward Syndrome was made, however, there was no karyotyping done to confirm the diagnosis. Discussion: Omphalocele with umbilical cord cyst is rare. Clinical providers should have a high suspicion of aneuploidy with the presence of multiple fetal anomalies in advanced maternal age. Prenatal detection and diagnosis are crucial to aid counselling with consideration of an early termination of pregnancy.

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

cyst; literature; omphalocele

EMTREE MEDICAL INDEX TERMS

abdominal wall defect; adult; aneuploidy; choroid plexus; conference abstract; counseling; diagnosis; Edwards syndrome; female; fetus; fetus echography; fetus malformation; heart left ventricle; human; karyotyping; liver; lung; male; maternal age; micrognathia; pregnancy; prognosis; straw; systematic review; thorax; umbilical cord; Wharton jelly

LANGUAGE OF ARTICLE

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LANGUAGE OF SUMMARY

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RECORD 21

TITLE

Prenatal detection of pure proximal 6q14.1 microduplication encompassing LCA5 gene: A variant of likely benign

AUTHOR NAMES

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ABSTRACT

Trisomy 6q is a recognizable syndrome which exhibits psychomotor/growth retardation, developmental/intellectual disabilities, feeding difficulties, facial dysmorphism, hearing loss, brain and heart malformations. The purpose of this study was to delineate the prenatal features of proximal 6q14.1 duplication in fetal period, which was rarely reported in clinic. Eight pregnant women who opted for amniocentesis due to the fetal ultrasound abnormalities, maternal serum screening or other indications for prenatal diagnosis between 2019 and 2020. Chromosomal microarray analysis and G-banding analysis were offered after informed consents were obtained. Cytogenetic prenatal investigation showed all fetuses presented normal karyotypes except case 4 exhibiting a balanced chromosomal translocation 46,XX,t (4;8)(p16;q24). The chromosomal microarray analysis detected 0.211-0.242 Mb duplications of 6q14.1 (chr6: 80109532-80351666, hg19) in all 8 cases, encompassing the morbid gene LCA5 in common. Seven pregnant women (P1-P7) continued their pregnancies and delivered healthy infants at term while the parents of case 8 opted for termination of pregnancy for severe abnormal ultrasound findings. Overall, all neonates were in a good healthy condition with no evident anomalies, ranging from 2 m to 16 m. It is proposed that 6q14.1 duplication involving LCA5 gene detected in our study might be variants of likely benign. However, further large-scale studies should be gathered to assess its pathogenicity. To our knowledge, our study is the first report focusing on prenatally detected proximal 6q14.1 duplication, accompanied by detailed clinic phenotypes. Diverse ultrasound findings were

observed in these cases, ranging from normal to abnormal. More evidence should be gathered to interpret the prenatal genotype-phenotype correlation of 6q14.1 duplication. For these cases with 6q14.1 microduplication, long term follow up should be carried out in case abnormal clinical symptoms or developmental-behavioral disorders emerge.

AUTHOR KEYWORDS

6q14.1 duplication
LCA5
prenatal diagnosis
variants of likely benign

EMTREE DRUG INDEX TERMS (MAJOR FOCUS)

Ica5 protein (endogenous compound); protein (endogenous compound)

EMTREE DRUG INDEX TERMS

genomic DNA (endogenous compound); unclassified drug

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

chromosome 6q14.1 microduplication (diagnosis); chromosome duplication (diagnosis); prenatal diagnosis

EMTREE MEDICAL INDEX TERMS

adult; amniocentesis; article; birth weight; blood analysis; body build; cerebral palsy; chromosome analysis; chromosome translocation; clinical article; clinical feature; cytogenetic analysis; cytogenetics; CytoScan 750K; dextrocardia; DNA purification kit; female; fetus; fetus echography; follow up; genetic counseling; genetic variability; genotype phenotype correlation; growth retardation; heart ventricle septum defect; human; human tissue; infant; inferior cava vein; karyotype; karyotype 46,XX; male; microarray analysis; newborn; nuchal translucency measurement; pathogenicity; pregnancy; pregnant woman; situs inversus; skeleton malformation; treatment indication; trisomy 21; umbilical cord

DEVICE TRADE NAMES

CytoScan 750K, United States

DEVICE MANUFACTURERS

(United States)Affymetrix
Qiagen

CAS REGISTRY NUMBERS

protein (67254-75-5)

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

Developmental Biology and Teratology (21)

Human Genetics (22)

Clinical and Experimental Biochemistry (29)

Pediatrics and Pediatric Surgery (7)

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RECORD 22

TITLE

Chromosomal Microarray Analysis in Fetuses Detected with Isolated Cardiovascular Malformation: A Multicenter Study, Systematic Review of the Literature and Meta-Analysis

AUTHOR NAMES

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MDPI

ABSTRACT

Cardiovascular malformations (CVM) represent the most common structural anomalies, occurring in 0.7% of live births. The CVM prenatal suspicion should prompt an accurate investigation with fetal echocardiography and the assessment through genetic counseling and testing. In particular, chromosomal microarray analysis (CMA) allows the identification of copy number variations. We performed a systematic review and meta-analysis of the literature, studying the incremental diagnostic yield of CMA in fetal isolated CVM, scoring yields for each category of heart disease, with the aim of guiding genetic counseling and prenatal management. At the same time, we report 59 fetuses with isolated CVM with normal karyotype who underwent CMA. The incremental CMA diagnostic yield in fetuses with isolated CVM was 5.79% (CI 5.54–6.04), with conotruncal malformations showing the higher detection rate (15.93%). The yields for ventricular septal defects and aberrant right subclavian artery were the lowest (2.64% and 0.66%). Other CVM ranged from 4.42% to 6.67%. In the retrospective cohort, the diagnostic yield was consistent with literature data, with an overall CMA diagnostic yield of 3.38%. CMA in the prenatal setting was confirmed as a valuable tool for investigating the causes of fetal cardiovascular malformations.

AUTHOR KEYWORDS

cardiovascular malformations

chromosomal microarray analysis

fetal malformations

genetic counseling

genetic testing
heart disease
prenatal diagnosis
structural anomalies

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

cardiovascular malformation; chromosome analysis; microarray analysis

EMTREE MEDICAL INDEX TERMS

diagnostic value; fetus; fetus malformation; genetic counseling; genetic screening; heart disease; heart ventricle septum defect; human; karyotype; karyotyping; Preferred Reporting Items for Systematic Reviews and Meta-Analyses; prenatal diagnosis; review; right subclavian artery; systematic review; vascular ring

EMBASE CLASSIFICATIONS

Cardiovascular Diseases and Cardiovascular Surgery (18)

Pediatrics and Pediatric Surgery (7)

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RECORD 23

TITLE

Guideline No. 427: Folic Acid and Multivitamin Supplementation for Prevention of Folic Acid–Sensitive Congenital Anomalies

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BOOK PUBLISHER

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ABSTRACT

Objective: To provide updated guidance on pre-conception folic acid and multivitamin supplementation for primary and secondary (recurrence) prevention of neural tube defects and other folate-sensitive congenital anomalies. **Target population:** Women aged 12–45 years who could become pregnant should be aware of the risk of serious birth defects without adequate pre-conception and first-trimester folic acid supplementation. **Options:** Optimizing folic acid supplementation is complex and depends on factors including dosage; type of supplement; bioavailability of folate from food, timing of initiating supplementation; and metabolic and genetic factors. For all women who could become pregnant, a low daily dosage of folic acid is recommended before conception and throughout pregnancy and breastfeeding. High-dosage folic acid supplementation is recommended only for women who can become pregnant and have had a previous pregnancy affected by a neural tube defect or other folate-sensitive congenital anomaly. Directed personalized approaches could be considered and adopted for women who can become pregnant and have complex risks (genetic, medical, or surgical risk factors), using new knowledge of co-factor metabolism and synergy, as well as red blood cell or serum folate testing. Such approaches would require changes to current provincial health care maternal serum folate screening/testing. **Outcomes:** New approaches to oral folic acid supplementation, including triage tools, need to be considered to optimize the benefits of decreasing risk of neural tube defects and folate-sensitive congenital anomalies. **Benefits, harms, and costs:** Oral folic acid supplementation, or dietary folate intake combined with a multivitamin/micronutrient supplement, is associated with lower rates of neural tube defects, other folate-sensitive birth defects, and obstetrical complications. The costs are those attributable to daily vitamin supplementation and a healthy, folate-rich diet. **Evidence:** A literature search was designed and carried in PubMed and the Cochrane Library databases from 1990 to 2021 using following MeSH terms and keywords (and variants): folic acid supplementation; folate food fortification; primary neural tube defect prevention; prevention of recurrence of neural tube defects; folate-sensitive birth defects; folate supplementation benefit; folate supplementation risk; folate pregnant woman

physiology; pregnant woman RBC folate level; pregnant woman serum folate levels; folate and epilepsy; folate and obesity. This guideline was based upon expert guidelines or opinions, systematic reviews, randomized controlled clinical trials, and observational case-control studies and case series retrieved, published in English from 1990 to 2021. Validation Methods: The authors rated the quality of evidence and strength of recommendations using the Grading of Recommendations Assessment, Development and Evaluation (GRADE) approach. See online Appendix A (Tables A1 for definitions and A2 for interpretations of strong and weak recommendations). Intended audience: Maternity health care providers (physicians, midwives, nurses) and other providers of pregnancy-related wellness and health counselling.

SUMMARY STATEMENTS: 1. Prevention of folate-sensitive anomalies should be evidence-based, and the benefits of preventing anomalies should balance any risks of folic acid supplementation (high). 2. Birth defects related to folate deficiency account for 2%–3% of prenatal or neonatal major anomalies and 4%–5% of total structural malformations or developmental conditions identified after birth. Folate-sensitive birth defects include neural tube defects, certain congenital heart and urinary tract defects, oral facial clefts, and limb-reduction anomalies (high).

RECOMMENDATIONS: 1. Any woman aged 12–45 years who can become pregnant should be advised by their health care provider to maintain a healthy, folate-rich diet and should undergo a brief periodic dietary review (strong, moderate). 2. Health care providers can consider promoting regular consumption of choline-rich foods (meat, egg yolk) during wellness visits (such as for birth control renewal, Pap testing, gynaecologic examination), whether or not the patient is contemplating pregnancy (strong, moderate). 3. Health care providers should advise all women aged 12–45 years considering or planning a pregnancy about the benefits of taking an oral daily multivitamin containing folic acid (0.4–1.0 mg) to optimize serum and red blood cell folate levels (strong, high). 4. Folic acid should be taken in a daily oral multivitamin that includes a 2.6-µg dose of vitamin B12 (strong, high). 5. Any woman aged 12–45 years who can become pregnant and has pre-conception obesity (body mass index >30.0 kg/m²) may require a more personal and focused assessment for folate supplementation to prevent fetal anomalies, such as a pre-conception fasting serum folate concentration assessment. If a woman with obesity has had a previous fetus or child with a folate-sensitive fetal anomaly other than a neural tube defect, she should

take a folic acid supplement containing the recommended dosage for women at increased risk (4–5 mg) (conditional, low). 6. High-dosage folate supplementation (oral dosage of 4–5 mg/d) should be used only for women at high risk; women who can become pregnant and who have had a previous pregnancy affected by a neural tube defect, have had a neural tube defect themselves, or have a first-degree relative with a neural tube defect (strong, moderate). 7. High-dosage supplementation requires 2 separate periods of supplementation: from pre-conception to 12 weeks gestation (see below), and from 12 weeks gestation until completion of breastfeeding, when the folic acid supplementation dosage reverts to the low-dosage regimen (strong, high). There are 2 options for supplementation in the first period: 1. Standard option: a total pre-conception oral daily dosage of 4 mg folic acid (1 oral multivitamin supplement that contains 1.0 mg of folic acid and 2.6 µg of vitamin B12, an iron supplement of 16–20 mg/d, and 3 1.0-mg folic acid tablets); (strong, high) or 2. Personalized option: requires the patient to first take an oral daily multivitamin containing folic acid (0.4–1.0 mg) and vitamin B12 within the first 4–6 weeks of a 3-month pre-conception period, then complete a blood test to determine her fasting serum folate level. A daily dosage of folic acid supplementation (from this pre-conception period until 12 weeks gestation) of 0.4–1.0 mg would be chosen if results were in the optimal range, and a daily dosage of more than 1.0 mg, if the results were sub-optimal (strong, moderate–high). 8. Moderate-dosage folate supplementation: Women who can become pregnant and have either an increased risk of having a fetus with an NTD or other folate-sensitive congenital anomaly or other medical-surgical conditions associated with a risk of folate deficiency require 2 separate periods of supplementation (strong, high). • From pre-conception to 12 weeks gestation, the supplementation dosage is 1.0 mg of folic acid daily (1 oral multivitamin supplement that contains 1.0 mg folic acid and 2.6 µg vitamin B12, and an iron supplement of 16–20 mg/d). • After 12 weeks gestation, the folic acid supplementation dosage reverts to the low-dosage regimen. 9. Low-dosage folate supplementation: Women who can become pregnant and are at low risk of having a fetus with an NTD or other folate-sensitive congenital anomaly should consume a pre-conception and first-trimester diet of folate-rich foods along with a daily oral multivitamin supplement that contains 0.4 mg (400 µg) of folic acid and 2.6 µg of vitamin B12, and an iron supplement of 16–20 mg daily for at least 2–3 months

before conception, throughout the pregnancy, and for 4–6 weeks postpartum or as long as breastfeeding continues (strong, high).

AUTHOR KEYWORDS

folate-sensitive anomalies

folic acid

multivitamins

neural tube defect

pregnancy

primary prevention

EMTREE DRUG INDEX TERMS (MAJOR FOCUS)

folic acid; multivitamin

EMTREE DRUG INDEX TERMS

choline; cyanocobalamin; trace element; vitamin B group

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

congenital heart malformation; neural tube defect; practice guideline; pregnancy; primary prevention

EMTREE MEDICAL INDEX TERMS

adolescent; adult; article; bioavailability; birth control; body mass; breast feeding; case control study; child; clinical trial; Cochrane Library; complication; conception; controlled study; counseling; diet; drug combination; drug dose regimen; drug megadose; drug therapy; egg yolk; epilepsy; erythrocyte; face; fasting; female; fetus; fetus malformation; first trimester pregnancy; first-degree relative; folate intake; folic acid blood level; folic acid deficiency; gynecological examination; health care personnel; heredity; human; human cell; human tissue; iron therapy; limb; low drug dose; maternal care; maternal serum; Medical Subject Headings; Medline; metabolism; midwife; newborn; nurse; obesity; oral drug administration; patient triage; physician; pregnant woman; prevention; randomized controlled trial (topic); school child; screening test; surgical risk; systematic review; urinary tract; vitamin blood level; vitamin supplementation

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RECORD 24**TITLE**

Prenatal diagnosis of a coronary-to-pulmonary artery fistula in a fetus with pulmonary atresia and ventricular septal defect

AUTHOR NAMES

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ABSTRACT

Pulmonary atresia and ventricular septal defect (PA/VSD) demonstrate a wide variety of pulmonary and coronary artery abnormalities; additionally, coronary-to-pulmonary artery fistula (CPAF) is a rare manifestation of PA/VSD and is seldom detected during pregnancy. In this report, we present a case of prenatal diagnosis of CPAF in PA/VSD and impactful images in a neonate, which were obtained using fetal echocardiography and postnatal electrocardiography-gated 320-row CT. Prenatal diagnosis of CPAF can facilitate the provision of better therapeutic strategies after birth.

AUTHOR KEYWORDS

Congenital disorders

Neonatal and paediatric intensive care

EMTREE DRUG INDEX TERMS

nitrogen (drug therapy, inhalational drug administration, special situation for pharmacovigilance)

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

coronary artery fistula (congenital disorder, diagnosis, surgery); heart ventricle septum defect (congenital disorder, diagnosis, surgery); prenatal diagnosis; pulmonary valve atresia (congenital disorder, diagnosis, surgery)

EMTREE MEDICAL INDEX TERMS

Aquilion ONE GENESIS Edition; article; blood vessel graft; cardiac conduit; cardiovascular patch; case report; clinical article; clinical decision making; color Doppler flowmetry; CT scanner; diagnostic accuracy; differential diagnosis; early intervention; echocardiograph; echocardiography; electrocardiography; fetus; fetus echography; gestational age; Gore Tex Graft; heart catheterization; heart left ventricle function; heart right ventricle function; heart right ventricle outflow tract; heart surgery; high flow nasal cannula therapy; human; male; newborn; parent counseling; primigravida; pulmonary artery stenosis (diagnosis); reconstructive surgery; shunting; tachypnea (diagnosis, drug therapy, therapy); transthoracic echocardiography; treatment duration; treatment planning; Volusion E8; x-ray computed tomography

DEVICE TRADE NAMES

Aquilion ONE GENESIS Edition

Gore Tex Graft

Volusion E8

DEVICE MANUFACTURERS

GE Healthcare

Gore

Toshiba Medical Systems

CAS REGISTRY NUMBERS

nitrogen (7727-37-9)

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

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RECORD 25**TITLE**

Molecular cytogenetic characterization of 16p11.2 microdeletions with diverse prenatal phenotypes: Four cases report and literature review

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ABSTRACT

Objective: Chromosome 16p11.2 deletions have been recognized as a genetic disorder with well-described postnatal phenotypes. However, the prenatal manifestations are atypical for lacking of enough evidence. Case report: Four pregnant women underwent amniocentesis for cytogenetic analysis and chromosomal microarray analysis (CMA) because of various indications for prenatal diagnosis: prenatal ultrasound abnormalities (cases 1, 2 and 4) and the childbearing history of cerebral palsy child (case 3). No overlapping phenotypes were observed in cases 1, 2 and 4, which might indicate phenotypic diversities in prenatal phenotypes for 16p11.2 microdeletion. All four fetuses showed normal karyotypic results while CMA identified 0.303–0.916 Mb microdeletions of 16p11.2, encompassing BP2–BP3 and BP4–BP5 regions separately. According to the parental CMA verification, case 1 carried a maternal inherited duplication in the region of Xp22.33 and a de novo deletion in the region of Xp21.1. All parents opted for the termination of pregnancies based upon genetic counselling. Conclusion: Our findings enriched the intrauterine phenotypic features of 16p11.2 microdeletions, which would be beneficial for genetic counselling in clinic. In addition, preimplantation genetic testing was recognized as a first-tier approach for such carriers if they intended to conceive again.

AUTHOR KEYWORDS

Chromosomal microarray analysis

Phenotypic diversities

Preimplantation genetic testing

Prenatal 16p11.2 microdeletion

EMTREE MEDICAL INDEX TERMS (MAJOR FOCUS)

chromosome deletion (diagnosis); cytogenetic analysis; cytogenetics; phenotype; prenatal diagnosis

EMTREE MEDICAL INDEX TERMS

adult; amniocentesis; article; case report; cerebral palsy; chromosomal instability; chromosome analysis; chromosome duplication; clinical article; female; fetus; fetus echography; genetic counseling; heart right ventricle dysplasia; heart ventricle septum defect; human; human cell; karyotype; microarray analysis; nasal bone; nuchal translucency measurement; pregnancy; pregnancy termination; right subclavian artery; tricuspid valve atresia; young adult

EMBASE CLASSIFICATIONS

Obstetrics and Gynecology (10)

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DOI

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EMBASE LINK

<https://www.embase.com/search/results?subaction=viewrecord&id=L2017718199&from=export>

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APÊNDICE C - RESULTADOS DE BUSCA NO LILACS

ID	Title	Authors	Source	Language / Publication year / Publication Country	Descriptor(s)	Fulltext URL	Entry Date	Volume number	Issue number	DOI	ISSN
biblio-1134392	Congenital Heart Disease Revealing Familial 22q11 Deletion Syndrome	Santos, Marlene Viviane Pires Fernandes; Gamba, Bruno Faulin; Empke, Stefany Lucas Lopes; Alves, Camila Cristina de Oliveira; Bérغامo, Nádia Aparecida; Ribeiro-Bicudo, Lucilene Arilho	Int. j. cardiovasc. sci. (Impr.);33(4): 425-426, July-Aug. 2020. tab, graf / Article	en / 2020 / BR	Humanos; Masculino; Feminino; Lactente; Pré-Escolar; Síndrome da Deleção 22q11/complicações; Cardiopatias Congênitas/genética; Fenótipo; Anormalidades Congênitas/genética; Insuficiência Velofaríngea; Síndrome de DiGeorge/genética; Aconselhamento Genético	http://www.scielo.br/scieop?script=sci_arttext&pid=S2359-5647202000400425	20201126	33	4	10.36660/ijcs.20180060	2359-4802
biblio-1128224	Síndrome de Edwards con cardiopatía congénita de larga supervivencia: reporte de caso y revisión de literatura/ Prolonged survival in Edwards syndrome	López-Ríos, Valery; Grajales-Marín, Estefanía; Gómez-Zambrano, Valentina; Barrios-Arroyave, Freddy Andrés	Medwave;20(8): e8015, 2020. / Article	en / 2020 / CL	Humanos; Feminino; Criança; Qualidade de Vida; Síndrome da Trissomia do Cromossomo 18/fisiopatologia; Cardiopatias Congênitas/fisiopatologia; Diagnóstico Pré-Natal; Ultrassonografia Pré-Natal; Síndrome da Trissomia do Cromossomo 18/diagnóstico; Síndrome da	https://www.medwave.cl/links/medwave/Estudios/Casos/8015.act	20201105	20	8	10.5867/medwave.2020.08.8015	0717-6384

	with congenital heart disease: a case report and literature review					Trissomia do Cromossomo 18/terapia; Cardiopatias Congênitas/diagnóstico; Cardiopatias Congênitas/terapia; Amniocentese						
bibliography-1055061	Pregnancy in Women with Complex Congenital Heart Disease. A Constant Challenge in Gravidy em Portadoras de Cardiopatias Congênitas Complexas: Um Constante Desafio	Avila, Walkiria Samuel; Ribeiro, Veronica Martins; Rossi, Eduardo Giusti; Binotto, Maria Angelica; Bortolotto, Maria Rita; Testa, Carolina; Francisco, Rossana; Hajjar, Ludhmilla Abraão; Miura, Nana	Arq. bras. cardiol; 113(6): 1062-1069, Dec. 2019. tab, graf / Article	en / 2019 / BR		Humanos; Feminino; Gravidez; Recém-Nascido; Adolescente; Adulto; Adulto Jovem; Complicações Cardiovasculares na Gravidez/fisiopatologia; Cardiopatias Congênitas/fisiopatologia; Complicações Cardiovasculares na Gravidez/classificação; Complicações Cardiovasculares na Gravidez/diagnóstico; Complicações Cardiovasculares na Gravidez/mortalidade; Prognóstico; Mortalidade Materna; Idade Gestacional; Mortalidade Fetal; Cardiopatias Congênitas/classificação; Cardiopatias Congênitas/diagnóstico; Cardiopatias Congênitas/mortalidade	http://www.scielo.br/scielo.php?script=sci_arttext&pid=S0066-782X20190197	20200325	113	6	10.5935/abc.20190197	0066-782X
bibliography-1016473	Primera experiencia en telemedicina dentro de un	Olivetti, M E; Macaron, S; Villa, A	Med. infant; 26(2): 151-155, Junio 2019.	es / 2019 / AR		Humanos; Gravidez; Recém-Nascido; Lactente; Pré-Escolar; Criança; Planos e Programas de	http://www.medicinainfantil.org.ar/	20190917	26	2		0328-0160

	programa nacional/ First experience in telemedicine within a national program		Tab, ilus / Article		Saúde; Telemedicina/instrumentação; Consulta Remota; Comunicação por Videoconferência/instrumentação; Cardiopatias Congênitas/diagnóstico; Argentina/epidemiologia	imag es/stories/volumen/2019/xxvi/2151.pdf					
biblio-1005962	Anomalia de Ebstein en diagnóstico prenatal/ Anomalia de Ebstein no diagnóstico pré-natal/ Ebstein's anomaly in prenatal diagnosis	Chibas Lamoth, Yurima; Marsillí Rivera, Arismelis; Sánchez Ramírez, Elsa	Rev. inf. cient;97(5): 1010-1019, 2018. ilus / Article	es / 2018 / CU	Humanos; Feminino; Gravidez; Diagnóstico Pré-Natal; Anomalia de Ebstein/diagnóstico por imagem; Cuidado Pré-Natal; Cardiopatias Congênitas	http://www.revinfcientificas.sld.cu/index.php/revinfcientificas/vol21/26/3883	20190710	97	5		1028-9933
biblio-1005109	Anomalia de Ebstein en diagnóstico prenatal/ Ebstein's anomaly in prenatal diagnosis	Chibas Lamoth, Yurima; Marsillí Rivera, Arismelis; Ramírez Sánchez, Elsa	Rev. inf. cient;97(3): 643-651, 2018. ilus / Article	es / 2018 / CU	Humanos; Feminino; Gravidez; Diagnóstico Pré-Natal; Anomalia de Ebstein/diagnóstico por imagem; Cuidado Pré-Natal; Cardiopatias Congênitas/diagnóstico	http://www.revinfcientificas.sld.cu/index.php/revinfcientificas/vol21/26/3766	20190628	97	3		1028-9933
biblio-899911	Diagnóstico prenatal de rhabdomioma fetal asociado a esclerosis tuberosa:	Gómez López, Enrique	Rev. chil. obstet. ginecol. (En línea); 82(3): 313-321, jun. 2017.	es / 2017 / CL	Humanos; Feminino; Gravidez; Adulto; Rhabdomioma/diagnóstico por imagem; Esclerose Tuberosa/diagnóstico por imagem; Doenças Fetais/diagnóstico por imagem;	http://www.scielo.cl/scielo.php?script=sci_arttext&pid=S07067175201700313	20170904	82	3	10.4067/s0717-75201700313	0717-7526

	propósito de un caso/ Prenatal diagnosis of fetal rhabdomyoma associated with tuberous sclerosis		tab, graf / Article		Neoplasias Cardíacas/diagnóstico por imagem; Rbdomioma/complicações; Esclerose Tuberosa/complicações; Ultrassonografia Pré-Natal; Neoplasias Cardíacas/complicações	17-75262017000300313						
lil-788914	Estenosis aórtica crítica y síndrome de corazón izquierdo hipoplásico de diagnóstico precoz en un hospital de segundo nivel/ Critical aortic stenosis and hypoplastic left heart syndrome of early diagnosis in a second-level hospital	Mejias Quintero, Mileidy Egleet; Román Barba, Violeta; Huertas González, José María; Salem Salem, Haidar; Galindo Izquierdo, Alberto	Rev. chil. obstet. ginecol.; 81(3): 229-233, jun. 2016. ilus / Article	es / 2016 / CL	Humanos; Feminino; Gravidez; Adulto; Ultrassonografia Pré-Natal; Síndrome do Coração Esquerdo Hipoplásico/diagnóstico por imagem; Estenose da Valva Aórtica/etiologia; Estenose da Valva Aórtica/diagnóstico por imagem; Síndrome do Coração Esquerdo Hipoplásico/complicações; Diagnóstico Precoce; Cardiopatias Congênitas	http://www.scielo.cl/scielo.php?script=sci_arttext&pid=S0717-7526201600030010	20160809	81	3	10.4067/S0717-7526201600030010	0048-766X	
lil-757074	Polidactilia, holoprosencefalia, labio y paladar hendido: no siempre es lo que parece/ Polydactily, holoprosencephaly,	Alvarado Socarras, Jorge Luis; Laverde Amaya, Diana Carolina; Prada, Carlos; García Carrillo, Johan	Arch. argent. pediatr.; 113(5): e290-e293, oct. 2015. tab, ilus / Article	es / 2015 / AR	Humanos; Masculino; Recém-Nascido; Trissomia/diagnóstico; Cromossomos Humanos Par 13; Macrossomia Fetal/diagnóstico; Deformidades Congênitas da Mão/diagnóstico; Holoprosencefalia /diagnóstico; Fenda	https://www.scielo.org.ar/doc/publicaciones/arg/2015/v113n5a23.pdf	20150904	113	5	10.5546/aap.2015.e290	0325-0075	

	cleft lip and cleft palate are not always what they seem: Case report				Labial/diagnóstico; Fissura Palatina/diagnóstico; Polidactilia/diagnóstico						
lil-746641	Characteristics of fetuses evaluated due to suspected anencephaly: a population-based cohort study in southern Brazil/ Características de fetos avaliados por suspeita de anencefalia: um estudo de coorte de base populacional do Sul do Brasil	Pelizzari, Emanuele; Valdez, Carolina Melendez; Picetti, Jamile dos Santos; Cunha, André Campos da; Dietrich, Cristine; Fell, Paulo Renato Krahl; Targa, Luciano Vieira; Zen, Paulo Ricardo Gazzola; Rosa, Rafael Fabiano Machado	São Paulo med. j;133(2): 101-108, Mar-Apr/2015. tab, graf / Article	en / 2015 / BR	Adolescente; Adulto; Feminino; Humanos; Recém-Nascido; Masculino; Gravidez; Adulto Jovem; Anencefalia; Feto/anormalidades; Aborto Legal/estatística & dados numéricos; Anencefalia/epidemiologia; Brasil/epidemiologia; Estudos de Coortes; Deficiência de Ácido Fólico; Morte Fetal/etiologia; Idade Gestacional; Idade Materna; Registros Médicos; Defeitos do Tubo Neural/epidemiologia; Defeitos do Tubo Neural; Morte Perinatal; Estudos Retrospectivos; Ultrassonografia Pré-Natal	http://www.scielo.br/scielo.php?script=sci_arttext&pid=S1516-31802015012608	20150522	133	2	10.1590/1516-31802015012608	15163180
biblio-871525	Investigação citogenômica tecidual post-mortem em portadores de malformações congênitas/	Dias, Alexandre Torchio	São Paulo; s.n; 2015. [123] p. ilustr., tab, graf. / Article	pt / 2015 / BR	Humanos; Masculino; Feminino; Recém-Nascido; Anormalidades Congênitas; Variações do Número de Cópias de DNA; Mortalidade Infantil; Reação em Cadeia da Polimerase	http://www.teses.usp.br/teses/disponiveis/5/5144/tde-06016-	20160121				

	Post-mortem tissue cytogenomics investigation in patients with congenital malformations				Multiplex; Patologia Molecular	111509/publico/AlexandreTorchioDias.pdf					
lil-6 81 96 2	Evolução perinatal e pediátrica de crianças com translucência nucal aumentada e cariótipo normal/ Perinatal and pediatric follow up of children with increased nuchal translucency and normal karyotype	Vieira, Lívia Adriano; Silva, Sônia Valadare s Lemos; Faria, Roberto Buenfil de; Lippi, Umberto Gazi; Lopes, Reginaldo Guedes Coelho	Rev. bras. ginecol. obstet ;35(6): 274-280, jun. 2013. ilus, tab / Article	pt / 2013 / BR	Adolescente; Adulto; Pré-Escolar; Feminino; Humanos; Lactente; Recém-Nascido; Pessoa de Meia-Idade; Gravidez; Adulto Jovem; Medição da Translucência Nucal; Seguidamentos; Cariótipo; Estudos Retrospectivos	http://www.scielo.br/scielo.php?script=sci_arttext&pid=S0100-7202013000600007	2013 0808	35	6	10.1590/S0100-7202013000600007	0100-7203
lil-6 52 20 1	Cardiopatias e gravidez/ Heart diseases and pregnancy	Lage, Eura Martins; Barbosa, Alexandre Simões	Feminina;40(1)jan.-fev. 2012. tab / Article	pt / 2012 / BR	Humanos; Feminino; Gravidez; Cardiopatias/complicações; Cardiopatias/etiologia; Complicações Cardiovasculares na Gravidez; Aconselhamento Genético; Arritmias Cardíacas/etiologia; Cardiopatia Reumática/epidemiologia; Cardiopatias Congênitas/epide	http://files.bvs.br/upload/S/0100-7254/2012/v40n1/a3079.pdf	2012 0911	40	1		0100-7254

					miologia; Insuficiência Cardíaca/etiologia ; Equipe de Assistência ao Paciente; Gravidez de Alto Risco; Cuidado Pré-Natal; Diagnóstico Pré-Natal; Atenção Terciária à Saúde						
lil-5 34 57 8	El embaraz o en la mujer con enfermed ad cardíaca congénita : enfoque para el cardiólog o/ Congenit al heart disease in the pregnant woman: a guide for the cardiologi st	Vanegas, Edgardo s; Urrea, Juan K	Rev. colom b. cardiol ;16(4): 170-1 77, jul.-ag o. 2009. / Article	es / 2009 / CO	Cardiopatas Congênitas; Gravidez	http:// www.sc .org .co/R EVIS TAS CC/v 16/v 16n4 /v16 n4/p df/v1 6n4a 5.pdf	2009 1111	16	4		01 20- 56 33
lil-4 26 86 7	Síndrome cardioesp lénico: presentac ión de casos clínicos/ Cardiospl enic syndrome : report of clinical cases	Quiroz V., Lorena; Muñoz S., Hernán; Parra C., Mauro; Alvarez Z., Patricia	Rev. chil. ultras on;8(4): 114-11 6, 2005. ilus, tab / Article	es / 2005 / CL	Adulto; Humanos; Feminino; Gravidez; Cardiopatas Congênitas; Doenças Fetais; Anormalidades Múltiplas; Feto/anormalidad es; Terceiro Trimestre da Gravidez; Síndrome; Ultrassonografia Pré-Natal		2006 0322	8	4		07 17- 36 95
lil-3 26 72 2	Estado actual del manejo del niño con cardiopatí a	Buendía Hernánd ez, Alfonso	Arch. cardiol . Méx;7 1(supl .1): S32-S	es / 2001 / MX	Cardiopatas Congênitas; Administração dos Cuidados ao Paciente/tendênci as; Cardiologia; Aconselhamento		2002 1007	71	supl .1		14 05- 99 40

	congénita / State of the art in congenital heart disease		35, ene.-mar. 2001. ilus / Article		Genético/tendências						
lil-310336	Técnica de evaluación de las venas pulmonares durante la exploración del corazón fetal/ Evaluación technique of the pulmonary y venous during fetal heart scanning	Viñals L., Fernando; Giuliano B., Arrigó	Rev. chil. obstet. ginecol;66(4): 317-322, 2001. ilus, tab / Article	es / 2001 / CL	Humanos; Adulto; Grávidas; Feminino; Coração Fetal; Veias Pulmonares; Ultrassonografia Pré-Natal; Cardiopatias Congênitas; Canal Arterial; Segundo Trimestre da Grávidas; Ultrassonografia Doppler em Cores	20020128	66	4			0048-766X
lil-320857	Obstetric and perinatal aspects in patients with congenital heart diseases	Oliveira, Tenilson Amaral; Avila, Walkiria Samuel; Grinberg, Max	São Paulo med. j;114(5): 1248-1254, Sep.-Oct. 1996. / Article	en / 1996 / BR	Humanos; Feminino; Grávidas; Recém-Nascido; Adolescente; Adulto; Cardiopatias Congênitas/cirurgia; Complicações Cardiovasculares na Grávidas/cirurgia; Resultado da Grávidas; Aborto Espontâneo/etiologia; Morte Fetal; Retardo do Crescimento Fetal; Seguidores; Recém-Nascido de Baixo Peso; Prognóstico	http://www.scielo.br/scielo.php?script=sci_arttext&pid=S1516-3181-1996-0050-0002	20021025	114	5		1516-3180
lil-89689	Doença cardíaca fetal: aspectos básicos	Cabral, Antonio Carlos Vieira; Rezende	Feminina;18(1): 14-5, jan.	pt / 1990 / BR	Grávidas; Humanos; Feminino; Aconselhamento Genético;	19900809	18	1			0100-7254

	do aconselhamento genético/ Fetal heart disease: basic aspects of the genetics	, Cezar Alencar de Lima	1990. tab / Article		Cardiopatas Congênitas							
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APÊNDICE D - RESULTADOS DE BUSCA NO SCIELO

ID	Title	Author(s)	Source	Journal	Language(s)	Publication year	Fulltext URL
S0066-782 X20090004 00010-scl	Deleção 22q11.2 em pacientes com defeito cardíaco conotruncal e fenótipo da síndrome da deleção 22q11.2	Belangero, Sintia Iole Nogueira, Bellucco, Fernanda T.S., Kulikowski, Leslie Domenici, Christofolini, Denise M., Cernach, Mirlene C. S. P., Melaragno, Maria Isabel	Arquivos Brasileiros de Cardiologia; 92(4); 307-311	Arquivos Brasileiros de Cardiologia	English, Spanish, Portuguese	2009	http://www.scielo.br/scielo.php?script=sci_artext&pid=S0066-782X200900040010&lang=en

APÊNDICE E - RESULTADOS DE BUSCA NO SCOPUS

Scopus-10-Analyze-Year	
Your query : ((ALL("Hearts, Fetal" or "Heart, Fetal" or "Fetal Hearts") AND ALL("Defect, Congenital Heart " or "Abnormality, Heart Heart"or "Abnormality Congenital Heart" or "Defect Heart, Malformation Of"or "Malformation Of Heart"or "Malformation Of Hearts" or "Defects, Congenital Heart" or "Heart Abnormalities" or "Heart Defect, Congenital" or "Congenital Heart Disease" or "Congenital Heart Diseases" or "Disease, Congenital Heart" or "Heart Disease, Congenital" or "Congenital Heart Defects") AND ALL("counseling" or "counseling, parent" or "parent counseling" or "guidance, parent" or "parent guidance"))) AND (LIMIT-TO (SUBJAREA,"MEDI") OR LIMIT-TO (SUBJAREA,"HEAL") OR LIMIT-TO (SUBJAREA,"NURS") OR LIMIT-TO (SUBJAREA,"MULT") OR LIMIT-TO (SUBJAREA,"PSYC") OR LIMIT-TO (SUBJAREA,"BIOC")))	
Number of results : 10	
YEAR	
2022	42
2021	43
2020	31
2019	37
2018	30
2017	22
2016	38
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2014	29
2013	21
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2000	7

1999	7
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1981	2

APÊNDICE F - RESULTADOS DE BUSCA NO WEB OF SCIENCE

@Article{ref1,
author={Holmes, K. W.
and Huang, J. H.
and Gutshall, K.
and Kim, A.
and Ronai, C.
and Madriago, E. J.},
title={Fetal counseling for congenital heart disease: is communication effective?},
journal={JOURNAL OF MATERNAL-FETAL & NEONATAL MEDICINE},
keywords={Congenital heart disease; fetal counseling},
abstract={Purpose To assess the accuracy of maternal understanding of fetal cardiac defects following initial fetal counseling. Methods Pregnant women with a fetal diagnosis of congenital heart disease (CHD) were surveyed regarding understanding of their fetus's heart defect. The survey asked: (1) for a description of the heart condition; (2) how confident they were in the diagnosis; (3) whether their fetus would require heart surgery. Two fetal cardiologists evaluated the maternal qualitative description. Partners were excluded from the study. Results Fifty-one participants consented and 39 completed the survey. Mean age was 31 years, 60{\%} had some college level or post-graduate education, 48{\%} had Medicaid insurance, and 81{\%} were Caucasian. More than three-quarters of participants, stated they had either "quite a bit" or "very much" understanding of their fetus's diagnosis. Maternal assessment matched the physician's assessment of accuracy with 77{\%} (N = 30) demonstrating either "quite a bit" or a "very accurate" description of the diagnosis. All women correctly understood if their fetus would require heart surgery. Highest level of maternal education positively correlated with the accuracy of diagnosis (regression coefficient 0.48, $p < .002$). However, confidence in the diagnosis was independent of both education (0.30, $p = .167$) and maternal age (-0.03, $p = .234$). Conclusions Fetal counseling is effective in conveying anatomy and the need for surgery; however, accuracy amongst women with lower levels of education and maternal confidence in understanding can be improved.},
note={APR 2021},
note={Times Cited in Web of Science Core Collection: 1 Total Times Cited: 1 Cited Reference Count: 7},
issn={1476-7058},
doi={10.1080/14767058.2021.1874909},
url={https://doi.org/10.1080/14767058.2021.1874909},
language={English}}

@Article{KovacevicAUGa,
author={Kovacevic, A.
and Elsasser, M.
and Fluhr, H.
and Muller, A.
and Starystach, S.
and Bar, S.
and Gorenflo, M.},
title={Counseling for fetal heart disease-current standards and best practice},
journal={TRANSLATIONAL PEDIATRICS},
year={AUG},

volume={10},
number={8},
pages={2225-2234},
keywords={Fetal cardiology; congenital heart disease (CHD); counseling; interdisciplinary approach; MATERNAL STRESS; NEURODEVELOPMENTAL DELAY; AORTIC VALVULOPLASTY; PRENATAL-DIAGNOSIS; PREGNANCY; INFORMATION; DEPRESSION; EXPERIENCE; STATEMENT; ANOMALIES},
abstract={Congenital heart disease (CHD) is the most common cause of major congenital anomalies affecting newborns. Prenatal detection of CHD has been improving continuously during the last two decades due to technical advances and thus optimized fetal cardiac imaging. Besides the in-utero diagnosis of CHD effective parental counseling is an integral part of any Fetal Cardiology Program. However, studies on the most effective techniques are scarce, as well as data on empirical assessment of counseling and its effectiveness. In this review article, we summarize current guidelines from different international associations and societies. We provide an updated literature overview evaluating current standards of counseling with regard to parental needs. This includes ethical aspects, counseling for univentricular disease and in-utero cardiac interventions. We discuss our method to assess counseling success for fetal heart defects by exploring different analytical dimensions that may be considered helpful in order to improve efficacy. Finally, we present a proposal of how to optimize a setting for counseling based on the current literature and our own data. In summary, parental counseling for fetal heart disease is complex and multidimensional. Significant expertise in fetal cardiology and physiology, potential progression of CHD, postnatal treatment strategies and knowledge of long-term sequelae is necessary. A structured approach, together with continuous improvement of communicative skills, may lead to more effective counseling for parents following a diagnosis of CHD in the fetus.},
note={AUG 2020},
note={Times Cited in Web of Science Core Collection: 3 Total Times Cited: 3 Cited Reference Count: 34},
issn={2224-4336},
language={English}

@Article{AllanDEC30,
author={Allan, L. D.
and Huggon, I. C.},
title={Counselling following a diagnosis of congenital heart disease},
journal={PRENATAL DIAGNOSIS},
year={DEC 30},
volume={24},
number={13},
pages={1136-1142},
keywords={counselling; fetus; congenital heart disease; OUTFLOW TRACT OBSTRUCTION; PRENATAL-DIAGNOSIS; GREAT-ARTERIES; SURGERY; TRANSPOSITION; TETRALOGY; FETUSES; INFANTS; FALLOT},
abstract={Counselling the parents following a diagnosis of fetal congenital heart disease (CHD) is as important a task for the fetal cardiologist, as the skill involved in achieving an accurate diagnosis. The counsellor will base prognosis not only on the diagnosis itself but also on the security of diagnosis, the stage in gestation and potential for change, the association with extracardiac malformations and the known results of treatment. Depending on the gestational age and legal situation the counsellor is operating in, termination of pregnancy may be one of the options to consider and one that should always be raised in discussion. Thus, the parents may be in the position of making a crucial decision concerning the management of the pregnancy on the basis of the information received, so it is vital

that the counsellor is truly able to communicate with them, whatever be their level of understanding.
Copyright (C) 2004 John Wiley Sons, Ltd.},
note={Times Cited in Web of Science Core Collection: 50 Total Times Cited: 57 Cited Reference
Count: 22},
issn={0197-3851},
doi={10.1002/pd.1071},
url={https://doi.org/10.1002/pd.1071},
language={English}

@Article{SwanMAY,
author={Swan, L.},
title={Congenital heart disease in pregnancy},
journal={BEST PRACTICE {\&} RESEARCH CLINICAL OBSTETRICS {\&} GYNAECOLOGY},
year={MAY},
volume={28},
number={4},
pages={495-506},
keywords={congenital heart disease; pregnancy; pre-conception counselling;
PULMONARY-HYPERTENSION; ADULTS; WOMEN; TETRALOGY; FALLOT; MANAGEMENT;
RECURRENCE; MORTALITY; OUTCOMES; DEFECTS},
abstract={The story of congenital heart disease is one of the major successes of medicine in the last
50 years. Heart conditions previously associated with early death are now successfully treated. Many
of these women are now in their child-bearing years wishing to have children of their own. All of these
women should be offered comprehensive pre-conception counselling by a dedicated multi-disciplinary
team. Each woman will present a unique set of cardiac and obstetric challenges that require an
individualised assessment of risk and a carefully documented care plan. In this chapter, I describe the
most common forms of congenital heart disease and the specific issues that should be assessed
before conception. I present a systematic approach to risk stratification and care planning. These
lesions range from mild disease with little implications for pregnancy to those with a sizable risk of
maternal mortality or complications. I will also discuss fetal risk factors. (C) 2014 Elsevier Ltd. All rights
reserved.},
note={Times Cited in Web of Science Core Collection: 18 Total Times Cited: 18 Cited Reference
Count: 38},
issn={1521-6934},
doi={10.1016/j.bpobgyn.2014.03.002},
url={https://doi.org/10.1016/j.bpobgyn.2014.03.002},
language={English}

@Article{LuNOV,
author={Lu, C. W.
and Wu, M. H.
and Wang, J. K.
and Lin, M. T.
and Chen, C. A.
and Chiu, S. N.
and Chiu, H. H.},
title={Preconception Counseling for Women with Congenital Heart Disease},
journal={ACTA CARDIOLOGICA SINICA},

year={NOV},
volume={31},
number={6},
pages={500-506},
keywords={Congenital heart disease; Pregnancy; SYSTEMIC RIGHT VENTRICLE; PREGNANCY OUTCOMES; CARDIAC OUTCOMES; TASK-FORCE; MANAGEMENT; TRANSPOSITION; GUIDELINES},
abstract={With advances that have been made over the recent decades in transcatheter and surgical interventions, most patients with congenital heart disease (CHD) can survive into adulthood. Overall, probably half of these surviving patients are female. When these female CHD patients reach childbearing age, however, pregnancy management will be a major issue. In order to meet the demands of fetal growth, the maternal cardiovascular system starts a series of adaptations beginning in early pregnancy. These adaptations include: decreased systemic and pulmonary vascular resistances, decreased blood pressure, expansion of the blood volume, increased heart rate and increased cardiac output. For women with CHD, this hemodynamic alteration may increase the risks of adverse cardiovascular events as well as the fetal and neonatal complications. Therefore, proper risk stratification and effective counseling for women with CHD who are planning their pregnancies is an important undertaking.},
note={Times Cited in Web of Science Core Collection: 3 Total Times Cited: 3 Cited Reference Count: 30},
issn={1011-6842},
doi={10.6515/ACS20150319B},
url={https://doi.org/10.6515/ACS20150319B},
language={English}

@Article{KovacevicSEP,
author={Kovacevic, A.
and Simmelbauer, A.
and Starystach, S.
and Elsasser, M.
and Sohn, C.
and Muller, A.
and Bar, S.
and Gorenflo, M.},
title={Assessment of Needs for Counseling After Prenatal Diagnosis of Congenital Heart Disease - A Multidisciplinary Approach},
journal={KLINISCHE PADIATRIE},
year={SEP},
volume={230},
number={5},
pages={251-256},
keywords={congenital heart disease; fetal cardiology; prenatal diagnosis; counseling; PARENTS; METAANALYSIS; MORBIDITY; CHILDREN},
abstract={Background Congenital heart disease is the most common cause of major congenital anomalies. After prenatal diagnosis effective counseling is crucial. However, little research has been undertaken in determining the most effective techniques. Objectives To develop a questionnaire suitable to assess parental needs for counseling. Material and Methods A questionnaire was developed by pediatric cardiologists, maternal-fetal-medicine specialists and sociologists. Likert scaled and open-ended questions are combined with socio-demographical data. The questionnaire was prospectively pilot-tested on 17 parents. We present first analyses of n =41 parents. Results Response

rate was 89.5 (%). The dependent variable "effective counseling" was measured in 5 dimensions (transfer of medical information, trust in medical staff, transparency of treatment process, coping resources and perceived situational control). The questionnaire's internal consistency is high (Cronbach's alpha > 0.7). First analyses show that 44.7 (%) perceived counseling as successful. Transfer of medical information seems difficult (36.6(%) success rate). Trust in medical staff was high with 75(%). Conclusions This newly developed tool measures counseling success in five dimensions. A multidisciplinary approach is recommended as methodological expertise is essential for constructing adequate tests. Preliminary data indicate that transfer of medical information is not easily achieved. Further analyses are needed to identify factors that determine counseling success.},
note={Times Cited in Web of Science Core Collection: 7 Total Times Cited: 7 Cited Reference Count: 17},
issn={0300-8630},
doi={10.1055/a-0633-3331},
url={https://doi.org/10.1055/a-0633-3331},
language={English}

@Article{NagataJAN,
author={Nagata, H.
and Yamamura, K.
and Matsuoka, R.
and Kato, K.
and Ohga, S.},
title={Transition in cardiology 2: Maternal and fetal congenital heart disease},
journal={PEDIATRICS INTERNATIONAL},
year={JAN},
volume={64},
number={1},
keywords={fetal congenital heart disease; maternal congenital heart disease; transitional care; GREAT-ARTERIES; PREGNANCY; WOMEN; POPULATION; DEFECTS; MANAGEMENT; TRANSPOSITION; CONTRACEPTION; MULTICENTER; PREVALENCE},
abstract={The number of women with congenital heart disease (CHD) reaching reproductive age has been increasing. Many women with CHDs are desirous of pregnancy, but they face issues regarding preconception, antepartum, and postpartum management. On the other hand, the fetal diagnosis of CHD has improved with advances in the technique and equipment for fetal echocardiography. Recently, experiences with fetal intervention have been reported in patients with severe CHD, such as critical aortic stenosis. Nevertheless, some types of CHD are challenge to diagnose prenatally, resulting in adverse outcomes. Medical care is part of the transitional care for women and fetuses with CHD during the perinatal period. Pre-conceptual and prenatal counseling play an important role in transitional care. Sex and reproductive education need to be performed as early as possible. We herein review the current status, important issues to be resolved, and the future of maternal and fetal CHD to relevant caregivers.},
note={e15098},
note={Times Cited in Web of Science Core Collection: 0 Total Times Cited: 0 Cited Reference Count: 53},
issn={1328-8067},
doi={10.1111/ped.15098},
url={https://doi.org/10.1111/ped.15098},
language={English}

@Article{KovacevicFEB26,
author={Kovacevic, A.
and Simmelbauer, A.
and Starystach, S.
and Elsasser, M.
and Muller, A.
and Bar, S.
and Gorenflo, M.},
title={Counseling for Prenatal Congenital Heart Disease-Recommendations Based on Empirical Assessment of Counseling Success},
journal={FRONTIERS IN PEDIATRICS},
year={FEB 26},
volume={8},
keywords={fetal cardiology; Congenital heart disease (CHD); counseling; empirical assessment; parental needs; NEURODEVELOPMENTAL DELAY; DIAGNOSIS; PARENTS; EXPERIENCES; INFORMATION; STATEMENT; WORLDWIDE},
abstract={Objectives: Empirical assessment of parental needs and affecting factors for counseling success after prenatal diagnosis of congenital heart disease (CHD). Methods:Counseling success after fetal diagnosis of CHD was assessed by a validated standardized questionnaire. The dependent variable "Effective Counseling" was measured in five created analytical dimensions (1. "Transfer of Medical Knowledge-ToMK"; 2. "Trust in Medical Staff-TiMS"; 3. "Transparency Regarding the Treatment Process-TrTP"; 4. "Coping Resources-CR"; 5. "Perceived Situational Control-PSC"). Analyses were conducted with regard to influencing factors and correlations. Results: Sixty-one individuals (n = 40 females, n = 21 males) were interviewed in a tertiary medical care center. Median gestational age at first parental counseling was 28 + 6 weeks. Parental counseling was performed four times (median), mostly by pediatric cardiologists (83.6%). Overall counseling was successful in 46.3%, satisfying in 51.9%, and unsuccessful in 1.9%. Analyses of the analytical dimensions show that counseling was less successful for TOMK (38.3%) and PSC (39%); success rates were higher if additional written information or links to web sources were provided (60 and 70%, respectively). Length of consultation was positively correlated to counseling success for ToMK (r = 0.458), TrTP (r = 0.636), PSC (r = 0.341), and TiMS (r = 0.501). Interruptions were negatively correlated to the dimensions TiMS (r = -0.263), and TrTP (r = -0.210). In the presence of high-risk CHD (37.5%) overall counseling success was lower (26.1%). By cross table analysis and to a low degree of positive correlation in one dimension (ToMK; r = 0.202), counseling tends to be less successful for ToMK, TrTP, and TiMS if parents have not been counseled by cardiologists. Analyses regarding premises show a parental need for a separate counseling room, which significantly impacts ToMK (r = -0,390) and overall counseling success (r = -0.333). A language barrier was associated with lower success rates for ToMK, TiMS, and CR (21.4, 42.9, and 30.8%). Conclusions: Data from this multidisciplinary study indicate that parents after fetal diagnosis of CHD need uninterrupted counseling of adequate duration and quality in a separate counseling room. Providing additional written information or links to adequate web sources after initial counseling seems necessary. High-risk CHD needs more attention for counseling. There is a trend towards more counseling success if provided by cardiologists.},
note={26},
note={Times Cited in Web of Science Core Collection: 9 Total Times Cited: 9 Cited Reference Count: 23},
issn={2296-2360},
doi={10.3389/fped.2020.00026},
url={https://doi.org/10.3389/fped.2020.00026},
language={English}}

@Article{MellanderDEC,
author={Mellander, M.},
title={Perinatal management, counselling and outcome of fetuses with congenital heart disease},
journal={SEMINARS IN FETAL & NEONATAL MEDICINE},
year={DEC},
volume={10},
number={6},
pages={586-593},
keywords={fetal heart; echocardiography; counselling; pregnancy outcome; COMPLETE ATRIOVENTRICULAR-BLOCK; INTACT VENTRICULAR SEPTUM; PULMONARY-ARTERY CONDUIT; SEVERE AORTIC-STENOSIS; PRENATAL-DIAGNOSIS; FETAL DIAGNOSIS; IN-UTERO; EXPERIENCE; IMPROVES; OPERATION},
abstract={Prenatal treatment options for fetal heart disease are still limited but pharmacological treatment of fetal tachyarrhythmias is usually effective. Prenatal catheter interventions are likely to be an option in selected fetal cardiac defects in the future. Delivery should be at a tertiary care centre if the need for immediate neonatal transport is anticipated. When a cardiac problem is diagnosed in a fetus, the parents should be counselled by a paediatric cardiologist specialized in fetal cardiology in close co-operation with the obstetric team. The rate of termination is influenced by gestational age at diagnosis, the severity of the heart defect and the presence of associated malformations. In fetuses with isolated cardiac malformations who are in sinus rhythm with good myocardial function and no or trivial atrioventricular valve regurgitation, the risk of spontaneous intra-uterine death is low. Prenatal echocardiography has the potential to improve postnatal survival in infants with critical heart defects, especially those with duct-dependent systemic or pulmonary circulations. (c) 2005 Elsevier Ltd. All rights reserved.},
note={Times Cited in Web of Science Core Collection: 19 Total Times Cited: 19 Cited Reference Count: 50},
issn={1744-165X},
doi={10.1016/j.siny.2005.08.002},
url={https://doi.org/10.1016/j.siny.2005.08.002},
language={English}}

@Article{Kohler2001,
author={Kohler, F.
and Fotuhi, P.
and Baumann, G.},
title={Pregnancy and congenital heart disease},
journal={ZEITSCHRIFT FUR KARDIOLOGIE},
year={2001},
volume={90},
pages={30-35},
keywords={pregnancy; congenital heart disease; recurrence risk; maternal risk; fetal risk; FAMILY-PLANNING REQUIREMENTS; BALLOON MITRAL VALVOTOMY; EISENMENGER-SYNDROME; FETAL; WOMEN; MANAGEMENT; OUTCOMES; DEFECTS; PATIENT; REPAIR},
abstract={With advanced diagnostic, and therapeutic techniques in pediatric cardiology and cardiac surgery, pregnancy can be an option for patients with congenital heart disease. A low overall maternal mortality and a healthy pregnancy require interdisciplinary cooperation between the cardiologist, obstetrician and general practitioner caring for the mother. Treatment and outcome will depend on the

type of cardiac malformation, on the functional impairment of the maternal heart (heart failure and/or cyanosis) and on the status of the fetus, with evidence of a better outcome for patients treated in specialized centers. However, even with recent advances in treatment, for women with primary pulmonary hypertension, Eisenmenger's syndrome, left heart obstruction or Marfan syndrome, pregnancy remains associated with a high maternal mortality. Therefore, these are conditions in which pregnancy is still absolutely contraindicated and a patient should be counselled to terminate the pregnancy. The risk of an inherited recurrence of a congenital heart disease is difficult to assess in an individual case because the majority of cardiac malformations are caused by multi-factored variables. But for some types of malformations (i.e., atrioventricular canal, Morbus Fallot) the incidence of cardiac malformation in the offspring of affected parents is slightly higher compared to the general population. Consequently, all patients with congenital heart disease should be offered genetic counselling and fetal echocardiography. In general, pregnancy in women with congenital heart disease has no significant long-term adverse effects and a second pregnancy is possible in the majority of the cases.},

note={Times Cited in Web of Science Core Collection: 7 Total Times Cited: 7 Cited Reference Count: 36},

issn={0300-5860},

language={German}

@Article{FredouilleSEP,

author={Fredouille, C.},

title={Diagnosis of fetal heart abnormality. Fetal heart},

journal={ANNALES DE PATHOLOGIE},

year={SEP},

volume={17},

number={4},

pages={300-305},

keywords={fetal heart; congenital heart disease; prenatal diagnosis; anatomy; classification},

abstract={To diagnose a fetal heart abnormality is one of the most important purpose of the foetopathologic examination performed after spontaneous abortion, intrauterine death or pregnancy termination. Frequency of cardiac abnormalities in fetal or maternal pathologies explains the significance of this diagnosis in a pertinent genetic counseling before next pregnancy. The diagnose of a fetal heart abnormality requires a good knowledge of: normal anatomy and examination technics of fetal heart, congenital heart abnormalities classification and knowledge of the most common diagnosis, diagnostic orientation according to the cardiopathy : fetal origin in genic syndrom, chromosomal abnormalities, malformative or sporadic association; maternal etiology (toxic, infection, metabolic). This procedure needs experience, acquired by rigourous practice and team working.},

note={Times Cited in Web of Science Core Collection: 6 Total Times Cited: 6 Cited Reference Count: 13},

issn={0242-6498},

language={French}

@Article{PeyvandiMAY-JUN,

author={Peyvandi, S.

and Rychik, J.

and Zhang, X. M.

and Shea, J. A.

and Goldmuntz, E.},

title={Preconceptual Folic Acid Use and Recurrence Risk Counseling for Congenital Heart Disease},
journal={CONGENITAL HEART DISEASE},
year={MAY},
month={JUN},
volume={10},
number={3},
pages={219-225},
keywords={Congenital Heart Disease; Recurrence Risk; Risk Factors; Folic Acid Supplementation;
CURRENT KNOWLEDGE; CARDIOVASCULAR DEFECTS; SCIENTIFIC STATEMENT;
SUPPLEMENT USE; MULTIVITAMIN; ADULTS; PREVALENCE; MANAGEMENT; PREGNANCY;
COMMITTEE},
abstract={Objective. Recurrence risk of congenital heart disease (CHD) in families with an affected first-degree relative is increased as compared with the general population. Advances in genetic testing and evidence that preconceptual folic acid supplementation may decrease risk of CHD warrant preventative counseling for at-risk families. Our goal was to document patterns of preconceptual folic acid supplementation and recurrence risk counseling in at-risk families in order to identify opportunities for improved preventative care. Design. Mothers referred for a fetal echocardiogram were prospectively enrolled. Cases were defined as mothers deemed to be at higher risk of having an affected fetus with CHD given an affected parent or affected previous pregnancy with CHD. Controls were defined as mothers with no prenatal risk factors. Mothers completed a validated questionnaire assessing use of folic acid supplementation and receipt of recurrence risk counseling. Chi-square analyses were performed to analyze questionnaire responses and demographic data. Results. A total of 314 subjects participated (controls = 216, cases = 98). Cases took preconceptual folic acid supplementation more often than controls ($P < .001$), but only 55\% started preconceptually. Maternal advanced education and counseling ($P < .001$) were associated with preconceptual supplementation, whereas complexity of CHD in the relative was not. While 70\% of cases received some recurrence risk counseling, those with advanced education and complex CHD in the affected relative were more likely to receive counseling. Few at-risk cases interacted with genetic services (19\%). Conclusions. At-risk mothers with lower education are less likely to take preconceptual folic acid supplementation or receive recurrence risk counseling. Health care providers should proactively provide this information to all at-risk patients and develop collaborations with genetic services.},
note={Times Cited in Web of Science Core Collection: 1 Total Times Cited: 1 Cited Reference Count: 30},
issn={1747-079X},
doi={10.1111/chd.12206},
url={https://doi.org/10.1111/chd.12206},
language={English}

@Article{IsonFEB,
author={Ison, H. E.
and Griffin, E. L.
and Parrott, A.
and Shikany, A. R.
and Meyers, L.
and Thomas, M. J.
and Syverson, E.
and Demo, E. M.
and Fitzgerald, K. K.
and Fitzgerald-Butt, S.
and Ziegler, K. L.

and Schartman, A. F.
and Stone, K. M.
and Helm, B. M.},
title={Genetic counseling for congenital heart disease - Practice resource of the national society of genetic counselors},
journal={JOURNAL OF GENETIC COUNSELING},
year={FEB},
volume={31},
number={1},
pages={9-33},
keywords={complex disease; congenital heart disease; genetic counseling; genetic testing; practice resource; HYPOPLASTIC LEFT-HEART; COMPARATIVE GENOMIC HYBRIDIZATION; EARLY DIABETIC PREGNANCY; COPY NUMBER VARIANTS; DE-NOVO MUTATIONS; CHROMOSOMAL MICROARRAY; CARDIOVASCULAR MALFORMATIONS; AMERICAN-COLLEGE; RISK-FACTORS; SCIENTIFIC STATEMENT},
abstract={Congenital heart disease (CHD) is an indication which spans multiple specialties across various genetic counseling practices. This practice resource aims to provide guidance on key considerations when approaching counseling for this particular indication while recognizing the rapidly changing landscape of knowledge within this domain. This resource was developed with consensus from a diverse group of certified genetic counselors utilizing literature relevant for CHD genetic counseling practice and is aimed at supporting genetic counselors who encounter this indication in their practice both pre- and postnatally.},
note={SEP 2021},
note={Times Cited in Web of Science Core Collection: 3 Total Times Cited: 3 Cited Reference Count: 103},
issn={1059-7700},
doi={10.1002/jgc4.1498},
url={https://doi.org/10.1002/jgc4.1498},
language={English}

@Article{CanobbioFEB21,
author={Canobbio, M. M.
and Warnes, C. A.
and Aboulhosn, J.
and Connolly, H. M.
and Khanna, A.
and Koos, B. J.
and Mital, S.
and Rose, C.
and Silversides, C.
and Stout, K.
and Council, Amer Heart Assoc
and Cardiology, Council Clinical
and Young, Council Cardiovasc Dis
and Genomics, Council Functional
and Res, Council Quality Care Outcomes},
title={Management of Pregnancy in Patients With Complex Congenital Heart Disease A Scientific Statement for Healthcare Professionals From the American Heart Association},
journal={CIRCULATION},
year={FEB 21},

volume={135},
number={8},
pages={E50-E87},
keywords={AHA Scientific Statements; heart defects, congenital; heart diseases; pregnancy;
MOLECULAR-WEIGHT HEPARIN; MATERNAL CARDIOVASCULAR DYNAMICS; GUIDELINES
WRITING COMMITTEE; ARTERIAL SWITCH OPERATION; SYSTEMIC RIGHT VENTRICLE;
CARDIAC-OUTPUT; CARDIOPULMONARY BYPASS; NATRIURETIC-PEPTIDE;
CESAREAN-SECTION; BLOOD-VOLUME},
abstract={Today, most female children born with congenital heart disease will reach childbearing age. For many women with complex congenital heart disease, carrying a pregnancy carries a moderate to high risk for both the mother and her fetus. Many such women, however, do not have access to adult congenital heart disease tertiary centers with experienced reproductive programs. Therefore, it is important that all practitioners who will be managing these women have current information not only on preconception counseling and diagnostic evaluation to determine maternal and fetal risk but also on how to manage them once they are pregnant and when to refer them to a regional center with expertise in pregnancy management.},
note={Times Cited in Web of Science Core Collection: 175 Total Times Cited: 191 Cited Reference Count: 225},
issn={0009-7322},
doi={10.1161/CIR.0000000000000458},
url={https://doi.org/10.1161/CIR.0000000000000458},
language={English}

@Article{KovacevicFEB,
author={Kovacevic, A.
and Bar, S.
and Starystach, S.
and Simmelbauer, A.
and Elsasser, M.
and Muller, A.
and Motlagh, A. M.
and Oberhoffer-Fritz, R.
and Ostermayer, E.
and Ewert, P.
and Gorenflo, M.
and Wacker-Gussmann, A.},
title={Objective Assessment of Counselling for Fetal Heart Defects: An Interdisciplinary Multicenter Study},
journal={JOURNAL OF CLINICAL MEDICINE},
year={FEB},
volume={9},
number={2},
keywords={fetal heart disease; antenatal diagnosis; counselling; interdisciplinary study; Pediatric Cardiology; Fetal Cardiology; Maternal-Fetal Medicine; Perinatology; sociology;
PRENATAL-DIAGNOSIS; NEURODEVELOPMENTAL DELAY; DISEASE; EXPERIENCES;
INFORMATION; STATEMENT; WORLDWIDE; PARENTS},
abstract={The objective of this study was to analyze parental counselling for fetal heart disease in an interdisciplinary and multicenter setting using a validated questionnaire covering medical, sociodemographic, and psychological aspects. n = 168 individuals were recruited from two pediatric heart centers and two obstetrics units. Overall, counselling was combined successful and satisfying in

>99%; only 0.7% of parents were dissatisfied. "Perceived situational control" was impaired in 22.6%. Adequate duration of counselling leads to more overall counselling success ($r = 0.368$ ***), as well as providing written or online information (57.7% vs. 41.5%), which is also correlated to more "Transfer of Medical Knowledge" ($r = 0.261$ ***). Interruptions of consultation are negatively correlated to overall counselling success ($r = -0.247$ **) and to "Transparency regarding the Treatment Process" ($r = -0.227$ **). Lacking a separate counselling room is associated with lower counselling success for "Transfer of Medical Knowledge" ($r = 0.210$ ***). High-risk congenital heart disease (CHD) is correlated to lower counselling success (42.7% vs. 71.4% in low-risk CHD). A lack of parental language skills leads to less overall counselling success. There is a trend towards more counselling success for "Transfer of Medical Knowledge" after being counselled solely by cardiologists in one center ($r = 0.208$). Our results indicate that a structured approach may lead to more counselling success in selected dimensions. For complex cardiac malformations, counselling by cardiologists is essential. Parental "Perceived Situational Control" is often impaired, highlighting the need for further support throughout the pregnancy.}

note={467},

note={Times Cited in Web of Science Core Collection: 4 Total Times Cited: 4 Cited Reference Count: 24},

issn={2077-0383},

doi={10.3390/jcm9020467},

url={https://doi.org/10.3390/jcm9020467},

language={English}

@Article{JoneJUN,

author={Jone, P. N.

and Schowengerdt, K. O.},

title={Prenatal Diagnosis of Congenital Heart Disease},

journal={PEDIATRIC CLINICS OF NORTH AMERICA},

year={JUN},

volume={56},

number={3},

pages={709-+},

keywords={Fetal echocardiography; Prenatal diagnosis; Neonatal heart disease; INCREASED NUCHAL TRANSLUCENCY; MAJOR CARDIAC DEFECTS; FETAL DIAGNOSIS; GREAT-ARTERIES; FETUSES; POPULATION; EXPERIENCE; TRANSPOSITION; OBSTRUCTION; COARCTATION},

abstract={This article presents advancements in the field of fetal echocardiography and the significant impact of these within the fields of pediatric cardiology, perinatology, and neonatology. A prenatal diagnosis of congenital heart disease allows for improved counseling of the parents, guides the timing and optimal location of delivery, and allows appropriate planning and consultation between the cardiologist and neonatologist. It also facilitates accurate diagnosis and management of fetal arrhythmias, identifies potential candidates for in utero cardiac intervention, and serves as the imaging guidance technique for these procedures. The goals, indications, advantages, limitations, and spectrum of congenital heart disease that can be diagnosed are reviewed.},

note={Times Cited in Web of Science Core Collection: 23 Total Times Cited: 27 Cited Reference Count: 26},

issn={0031-3955},

doi={10.1016/j.pcl.2009.04.002},

url={https://doi.org/10.1016/j.pcl.2009.04.002},

language={English}

@Article{EvansMAR,
author={Evans, W. N.
and Acherman, R. J.
and Restrepo, H.},
title={Critical congenital heart disease and maternal comorbidities: An observation},
journal={PROGRESS IN PEDIATRIC CARDIOLOGY},
year={MAR},
volume={64},
keywords={Congenital heart disease; Fetal echocardiography; Prenatal diagnosis; Maternal comorbidities; PRENATAL DETECTION; SOUTHERN NEVADA; ASSOCIATION; DEFECTS},
abstract={Background: Critical congenital heart disease is the target of postnatal, pulse-oximetry screening; nevertheless, prenatal detection is preferable. Prenatal diagnosis allows for advanced perinatal planning, parental counseling, potential fetal cardiac intervention if indicated, and avoidance of complicated, costly, and at times, risky neonatal transport. Objective: The purpose of this study was to investigate a possible association between the occurrence of critical congenital heart disease and pregnancies complicated by maternal comorbidities. Methods: We identified all those prenatally and postnatally diagnosed with critical congenital heart disease in Nevada between July 2014 and June 2020. For each identified case of critical congenital heart disease, we assessed each mother's medical history. We divided pregnant women into two groups: 1) those with maternal comorbidities and 2) those without comorbidities. Results: We identified 367 cases of critical congenital heart disease. Of the 367, 229 (62\{\%\}) occurred in those with maternal comorbidities. Of the 367, 321 (87\{\%\}) were live-born. Of the 321 live-born, 201 (63\{\%\}) occurred in those with maternal comorbidities, however, the prevalence of maternal comorbidities in the general pregnancy population is not known. The majority of patients born with critical heart disease were born to mothers with comorbidities versus those without comorbidities for both the total and live-born cohorts ($p = 0.00001$). Of the 321 live-born, 10 had no prenatal care. Of the 311 with prenatal care, 248 (80\{\%\}) were prenatally diagnosed. During the study period, there were approximately 210,000 live births in Nevada, for a prevalence of 15.2 (321/210,000) critical congenital cardiac malformations per 10,000 live births. Conclusion: In Nevada, most of those with critical congenital heart disease are products of pregnancies complicated by maternal comorbidities. Further, the state-wide, live-born, critical congenital heart disease prenatal detection rate was 80\{\%\}.},
note={101433},
note={Times Cited in Web of Science Core Collection: 1 Total Times Cited: 1 Cited Reference Count: 14},
issn={1058-9813},
doi={10.1016/j.ppedcard.2021.101433},
url={https://doi.org/10.1016/j.ppedcard.2021.101433},
language={English}}

@Article{WarnesJUL,
author={Warnes, C. A.},
title={Pregnancy and Delivery in Women With Congenital Heart Disease},
journal={CIRCULATION JOURNAL},
year={JUL},
volume={79},
number={7},
pages={1416-1421},

keywords={Congenital heart disease; Eisenmenger syndrome; Genetic counseling; Preconception counseling; Pregnancy; MUSTARD OPERATION; OUTCOMES; ANTICOAGULATION; TRANSPOSITION; MANAGEMENT; VALVES; FETAL; COMPLICATIONS; WARFARIN; IMPACT},
abstract={Because of the growing population of patients with congenital heart disease (CHD), most maternal cardiac disease is now congenital in origin. For women with complex CHD, pregnancy poses an increased risk for both the mother, with complications of arrhythmias and heart failure being the most common, and the baby, with a higher chance of miscarriage, intrauterine growth retardation, and the need for early delivery. Pre-pregnancy counseling must be performed by cardiologists who have expertise in both CHD and pregnancy, with a detailed clinical assessment of the patient and the current hemodynamic situation, including echocardiography and an exercise test. In each case the approach must be individualized with consideration of the risks in each case. In some cases, such as Eisenmenger syndrome, pregnancy is contraindicated. Optimum outcomes in these complex patients are achieved when a multidisciplinary approach is used, involving maternal-fetal medicine specialists, cardiologists with expertise in CHD and obstetric anesthesia.},
note={Times Cited in Web of Science Core Collection: 29 Total Times Cited: 31 Cited Reference Count: 36},
issn={1346-9843},
doi={10.1253/circj.CJ-15-0572},
url={https://doi.org/10.1253/circj.CJ-15-0572},
language={English}

@Article{JainMAR-APR,

author={Jain, V. D.

and Moghbeli, N.

and Webb, G.

and Srinivas, S. K.

and Elovitz, M. A.

and Pare, E.},

title={Pregnancy in Women with Congenital Heart Disease: The Impact of a Systemic Right Ventricle},

journal={CONGENITAL HEART DISEASE},

year={MAR},

month={APR},

volume={6},

number={2},

pages={147-156},

keywords={Pregnancy; Maternal Heart Disease; Systemic Right Ventricle; Congenital Heart Disease; CORRECTED TRANSPOSITION; ATRIAL REPAIR; GREAT; OUTCOMES; DYSFUNCTION; SINGLE},

abstract={Objective. Individuals with a systemic right ventricle develop cardiac complications earlier in life. Limited data exists regarding the effect of a maternal systemic right ventricle on cardiac events during pregnancy. We sought to assess the effect of a systemic right ventricle on cardiac events and pregnancy outcomes. Design. The study was designed as a retrospective cohort study of pregnant women with maternal congenital heart disease. Setting. The study was set in a university, academic tertiary care referral center. Patients. Study subjects were identified by International Statistical Classification of Diseases and Related Health Problems-9 codes. Women with mitral valve prolapse only or noncongenital cardiac disease were excluded. The exposure was defined by systemic ventricle. Outcome Measures. The primary outcome was a composite of congestive heart failure, arrhythmia, stroke, cardiac arrest/death during pregnancy or postpartum (CARDCOMP). The secondary outcome (PREGCOMP) was a composite of preterm delivery, preeclampsia, growth restriction, and stillbirth/pregnancy loss (PREGCOMP). Student's t-test or chi-square/Fisher's exact

tests were used for comparison of continuous/categorical variables. Multivariable logistic regression was performed to control for possible confounders. Results. One hundred forty-six pregnancies in 114 women were included; 15 (10.3%) pregnancies involved a systemic right ventricle. CARDCOMP complicated 12.3% of these pregnancies. Women with a systemic right ventricle were more likely to develop CARDCOMP even after adjustment for confounders (odds ratio [OR] 6.32 [1.7-23.5], P = .006). PREGCOMP complicated 40.4% of all pregnancies. Women with a systemic right ventricle were also more likely to develop PREGCOMP (OR 5.37 [1.4-20.7], P = .015) compared with women with a systemic left ventricle after controlling for confounders. Conclusion. In women with congenital heart disease, a systemic right ventricle is associated with adverse cardiac and pregnancy outcomes. This information is critical for counseling and caring for these women. Further investigation is warranted regarding the effect of pregnancy on long-term health for this unique cohort of women.},
note={Times Cited in Web of Science Core Collection: 15 Total Times Cited: 15 Cited Reference Count: 26},
issn={1747-079X},
doi={10.1111/j.1747-0803.2011.00497.x},
url={https://doi.org/10.1111/j.1747-0803.2011.00497.x},
language={English}

@Article{DonofrioMAY,
author={Donofrio, M. T.},
title={Predicting the Future: Delivery Room Planning of Congenital Heart Disease Diagnosed by Fetal Echocardiography},
journal={AMERICAN JOURNAL OF PERINATOLOGY},
year={MAY},
volume={35},
number={6},
pages={549-552},
keywords={fetal echocardiography; prenatal diagnosis; delivery management; congenital heart disease; MATERNAL HYPEROXYGENATION; CARDIAC INTERVENTION; IN-UTERO; FETUSES; OUTCOMES; UTILITY; RISK; CARE},
abstract={Advances in prenatal imaging have improved the examination of the fetal cardiovascular system. Fetal echocardiography facilitates the prenatal diagnosis of congenital heart disease (CHD) and through sequential examination, allows assessment of fetal cardiac hemodynamics, predicting the evolution of anatomical and functional cardiovascular abnormalities in utero and during the transition to a postnatal circulation at delivery. This approach allows detailed diagnosis with prenatal counseling and enables planning to define perinatal management, selecting the fetuses at a risk of postnatal hemodynamic instability who are likely to require a specialized delivery plan. The prenatal diagnosis and management of critical neonatal CHD has been shown to play an important role in improving the outcome of newborns with these conditions, allowing timely stabilization of the circulation prior to cardiac intervention or surgery, thus reducing the risk of perioperative morbidity and mortality. Diagnostic protocols aimed at risk-stratifying severity and potential postnatal compromise in fetuses with CHD have been developed to identify those who may require special intervention at birth or within the first days of life. In addition, new methodologies are being studied to improve the accuracy of prediction of disease severity. Perinatal management of neonates with a prenatal diagnosis of CHD requires a close collaboration between obstetric, neonatal, and cardiology services. In this article, the management of fetuses with CHD will be discussed, along with summarizing the in utero and fetal echocardiographic findings used for risk stratification of newborns with CHD and reviewing the basic principles used for planning for neonatal resuscitation and initial transitional care of these complex newborns.},

note={Times Cited in Web of Science Core Collection: 10 Total Times Cited: 13 Cited Reference Count: 17},
issn={0735-1631},
doi={10.1055/s-0038-1637764},
url={https://doi.org/10.1055/s-0038-1637764},
language={English}

@Article{WielandnerOCT,
author={Wielandner, A.
and Mlczoch, E.
and Prayer, D.
and Berger-Kulemann, V.},
title={Potential of magnetic resonance for imaging the fetal heart},
journal={SEMINARS IN FETAL & NEONATAL MEDICINE},
year={OCT},
volume={18},
number={5},
pages={286-297},
keywords={Congenital heart disease; Fetal heart; Magnetic resonance imaging;
PRENATAL-DIAGNOSIS; CARDIAC ABNORMALITIES; CLINICAL-APPLICATIONS; DISEASE; MRI;
FEASIBILITY; COARCTATION; ULTRASOUND; SURVIVAL; INFANTS},
abstract={Significant congenital heart disease (sCHD) affects 3.6 per 1000 births, and is often associated with extracardiac and chromosomal anomalies. Although early mortality has been substantially reduced and the rate of long-term survival has improved, sCHD is, after preterm birth, the second most frequent cause of neonatal infant death. The prenatal detection of cardiac and vascular abnormalities enables optimal parental counselling and perinatal management. Echocardiography (ECG) is the first-line examination and gold standard by which cardiac malformations are defined. However, adequate examination by an experienced healthcare provider with modern technical imaging equipment is required. In addition, maternal factors and the gestational age may lower the image quality. Fetal magnetic resonance imaging (MRI) has been implemented over the last several years and is already used in the clinical routine as a second-line approach to assess fetal abnormalities. MRI of the fetal heart is still not routinely performed. Nevertheless, fetal cardiac MRI has the potential to complement ultrasound in detecting cardiovascular malformations and extracardiac lesions. The present work reviews the potential of MRI to delineate the anatomy and pathologies of the fetal heart. This work also deals with the limitations and continuing developments designed to overcome the current problems in cardiac imaging, including fast fetal heart rates, the lack of ECG-gating, and the presence of fetal movements. (C) 2013 Elsevier Ltd. All rights reserved.},
note={Times Cited in Web of Science Core Collection: 22 Total Times Cited: 24 Cited Reference Count: 44},
issn={1744-165X},
doi={10.1016/j.siny.2013.05.006},
url={https://doi.org/10.1016/j.siny.2013.05.006},
language={English}

@Article{GedikbasiMAR,
author={Gedikbasi, A.
and Oztarhan, K.
and Yildirim, G.

and Gul, A.
and Ceylan, Y.},
title={Counseling and outcomes of antenatally diagnosed congenital heart anomalies in Turkey},
journal={ANATOLIAN JOURNAL OF CARDIOLOGY},
year={MAR},
volume={11},
number={2},
pages={137-145},
keywords={Congenital heart defects; counseling; outcome; extracardiac malformations; ultrasound;
fetal echocardiography; PRENATAL-DIAGNOSIS; DISEASE; BIRTH; MALFORMATIONS;
POPULATION; PREVALENCE; EXPERIENCE; DEFECTS},
abstract={Objective: To determine the clinical outcomes and decisions of families of fetuses with
prenatally-diagnosed cardiac abnormalities. Methods: Prenatally diagnosed cases (n=155) with
congenital heart disease were retrospectively categorized according to the Allan-Huggon grading
system: Group A (cardiac disease associated with severe / lethal extracardiac disease); Group B1 (low
risk with a postnatal prognosis); Group B2 (moderate risk, amenable to surgical repair with a low
mortality); and Group B3 (high risk, associated with high mortality after surgery). Neonatal outcomes,
including termination of pregnancy, were recorded for 18 months of follow-up after counseling the
parents. Student's t-test, Mann-Whitney U, Pearson's Chi-square test and Fischer's exact Chi-square
test were used for statistical analyses. Results: One hundred forty-five cases completed follow up.
Thirty-nine cases (Group A) were associated with extracardiac lethal defects and the pregnancies
were terminated; these cases were excluded from statistical evaluation. Twenty parents in Group B3
opted also for termination. The survival rates of ongoing pregnancies after 18 months of follow-up
between the three cardiac abnormality Groups (Group B1, n=37; Group B2, n=12; and Group B3,
n=37) were 89.2{\\%}, 66.7{\\%}, and 13.5{\\%}, respectively. Significance was present between the
survival rates of the three Groups [Group B3 vs. Group B1: p=0.0001; OR: 52.8 (12.9-214.5); Group
B3 vs. Group B2: p=0.0009; OR: 12.8 (2.8-58.9); Group B2 vs. Group B1: p=0.087; OR: 4.12
(0.84-20.2)] Conclusion: Our practice and the findings reported herein support the efficacy of this
staging system and counseling parents of fetuses for congenital heart diseases. (Anadolu Kardiyol
Berg 2011; 11: 137-45)},
note={Times Cited in Web of Science Core Collection: 5 Total Times Cited: 6 Cited Reference Count:
25},
issn={2149-2263},
doi={10.5152/akd.2011.035},
url={https://doi.org/10.5152/akd.2011.035},
language={English}

@Article{KillenOCT,
author={Killen, S. A. S.
and Mouledoux, J. H.
and Kavanaugh-McHugh, A.},
title={Pediatric prenatal diagnosis of congenital heart disease},
journal={CURRENT OPINION IN PEDIATRICS},
year={OCT},
volume={26},
number={5},
pages={536-545},
keywords={congenital heart disease; fetal arrhythmias; prenatal diagnosis; LONG QT SYNDROME;
FETAL ECHOCARDIOGRAPHY; PULMONARY-ATRESIA; GREAT-ARTERIES;

ATRIOVENTRICULAR-BLOCK; CLINICAL PRESENTATION; ANTENATAL DIAGNOSIS; TRAINING-PROGRAM; GESTATIONAL-AGE; 4-CHAMBER VIEW},
abstract={Purpose of review Fetal cardiology is a rapidly evolving field. Imaging technology continues to advance as do approaches to in-utero interventions and care of the critically ill neonate, with even greater demand for improvement in prenatal diagnosis of congenital heart disease (CHD) and arrhythmias. Recent findings Reviewing the advances in prenatal diagnosis of CHD in such a rapidly developing field is a broad topic. Therefore, we have chosen to focus this review of recent literature on challenges in prenatal detection of CHD, challenges in prenatal counseling, advances in fetal arrhythmia diagnosis, and potential benefits to patients with CHD who are identified prenatally. Summary As methods and tools to diagnose and manage CHD and arrhythmias in utero continue to improve, future generations will hopefully see a reduction in both prenatal and neonatal morbidity and mortality. Prenatal diagnosis can and should be used to optimize location and timing of delivery and postnatal interventions. Video abstract <http://links.lww.com/MOP/A21>},
note={Times Cited in Web of Science Core Collection: 9 Total Times Cited: 10 Cited Reference Count: 98},
issn={1040-8703},
doi={10.1097/MOP.0000000000000136},
url={<https://doi.org/10.1097/MOP.0000000000000136>},
language={English}

@Article{WimalasunderaDEC30,
author={Wimalasundera, R. C.
and Gardiner, H. M.},
title={Congenital heart disease and aneuploidy},
journal={PRENATAL DIAGNOSIS},
year={DEC 30},
volume={24},
number={13},
pages={1116-1122},
keywords={congenital heart disease; aneuploidy; karyotype; 22q11; trisomy 21; HYPOPLASTIC LEFT-HEART; ATRIOVENTRICULAR SEPTAL-DEFECTS; CHROMOSOME 22Q11 DELETIONS; PRENATAL-DIAGNOSIS; DOWN-SYNDROME; CARDIAC-MALFORMATIONS; FETAL ECHOCARDIOGRAPHY; NUCHAL TRANSLUCENCY; DIGEORGE-SYNDROME; GENETIC ETIOLOGY},
abstract={Congenital heart disease (CHD) is one of the commonest prenatal diagnoses made on routine ultrasound screening. Overall, up to 33{\%} of CHD are associated with fetal aneuploidy. However, some specific cardiac lesions have a significantly greater association with particular chromosomal abnormalities. The majority of fetuses with CHD and aneuploidy also have extra-cardiac anomalies and are best managed by a multidisciplinary team where the management and prognosis of the cardiac defect can be discussed in the context of the baby as a whole. It is therefore important for clinicians involved in the management of fetuses with CHD to be aware of the association of aneuploidy as well as the prognosis and management of these cases, so that they can appropriately counsel the parents. In this chapter, we review the frequency and types of aneuploidy associated with the commonly diagnosed CHD and discuss their management. Copyright (C) 2004 John Wiley Sons, Ltd.},
note={Times Cited in Web of Science Core Collection: 44 Total Times Cited: 48 Cited Reference Count: 61},
issn={0197-3851},
doi={10.1002/pd.1068},
url={<https://doi.org/10.1002/pd.1068>},

language={English}

@Article{KovacevicJAN,
author={Kovacevic, A.
and Wacker-Gussmann, A.
and Bar, S.
and Elsasser, M.
and Motlagh, A. M.
and Ostermayer, E.
and Oberhoffer-Fritz, R.
and Ewert, P.
and Gorenflo, M.
and Starystach, S.},
title={Parents' Perspectives on Counseling for Fetal Heart Disease: What Matters Most?},
journal={JOURNAL OF CLINICAL MEDICINE},
year={JAN},
volume={11},
number={1},
keywords={fetal cardiology; parental counseling; social science; parental needs;
PRENATAL-DIAGNOSIS; MATERNAL STRESS; BAD-NEWS; PREGNANCY; INFORMATION;
DEPRESSION},
abstract={After diagnosis of congenital heart disease (CHD) in the fetus, effective counseling is considered mandatory. We sought to investigate which factors, including parental social variables, significantly affect counseling outcome. A total of n = 226 parents were recruited prospectively from four national tertiary medical care centers. A validated questionnaire was used to measure counseling success and the effects of modifiers. Multiple linear regression was used to assess the data. Parental perception of interpersonal support by the physician (beta = 0.616 ***, p = 0.000), counseling in easy-to-understand terms (beta = 0.249 ***, p = 0.000), and a short period of time between suspicion of fetal CHD, seeing a specialist and subsequent counseling (beta = 0.135 **, p = 0.006) significantly improve "overall counseling success". Additional modifiers (e.g., parental native language and age) influence certain subdimensions of counseling such as "trust in medical staff" (language effect: beta = 0.131 *, p = 0.011) or "perceived situational control" (age effect: beta = 0.166 *, p = 0.010). This study identifies independent factors that significantly affect counseling outcome overall and its subdimensions. In combination with existing recommendations our findings may contribute to more effective parental counseling. We further conclude that implementing communication skills training for specialists should be considered essential.},
note={278},
note={Times Cited in Web of Science Core Collection: 1 Total Times Cited: 1 Cited Reference Count: 31},
issn={2077-0383},
doi={10.3390/jcm11010278},
url={https://doi.org/10.3390/jcm11010278},
language={English}

@Article{RakhaJUL-AUG,
author={Rakha, S.},
title={Awareness assessment for parents of children with congenital heart diseases regarding fetal echocardiography},

journal={TURKISH JOURNAL OF PEDIATRICS},
year={JUL},
month={AUG},
volume={62},
number={4},
pages={569-577},
keywords={awareness; parents; congenital heart disease; fetal echocardiography; SCIENTIFIC STATEMENT; DIAGNOSIS; RECURRENCE; DEFECTS; ASSOCIATION; PREGNANCIES},
abstract={Background and objectives. The high recurrence rate of congenital heart disease (CHD) in siblings was the rationale for recommending fetal echocardiography. However, in a developing country like Egypt, there are limited fetal echocardiographic examinations under this category. The study was conducted to assess knowledge about fetal echocardiography amongst parents of children with CHD. Method. A questionnaire survey was conducted in a tertiary pediatric hospital from June to December 2018. The study included parents having children with CHD follow up in the outpatient clinic or admitted in the pediatric cardiology ward. The questionnaire included demographic data of parents regarding sex, age, education, and residence. It also includes a detailed inquiry about parents' knowledge regarding the availability, safety, and value of fetal echocardiography. Results. Participants were 200 parents, mostly mothers 159 (79.5%). The median age of parents included in the study was 33.5 (29-40) years. Regarding awareness, 134 (67%) did not know any prenatal diagnostic investigation for CHD, 46 (23%) knew fetal echocardiography, and 20 (10%) named other tools. Nevertheless, 34% of parents thought they would need fetal echocardiography in a subsequent pregnancy. Although 178 (89%) of parents thought it might be a safe investigation, 33% did not think it will have additional benefits over postnatal echocardiography. The age of the patient and age of the participating parent were the only statistically significant predictors for parents knowledge on fetal echocardiography existence with $p=0.008$, 95% CI=1.0391.282 and $p=0.015$, 95% CI = 0.864-0.984, respectively. Conclusion. Parents of children with CHD have significant knowledge gaps regarding Fetal Echocardiography. Our findings suggest that the current parent counseling is inadequate and needs further focus, especially in developing countries, to promote parents' understanding of the prenatal cardiac diagnosis. Data on fetal echocardiography should be clarified at the initial diagnosis of pediatric CHD.},
note={Times Cited in Web of Science Core Collection: 0 Total Times Cited: 0 Cited Reference Count: 21},
issn={0041-4301},
language={English}

@Article{ParriniAUG,
author={Parrini, I.
and Luca, F.
and Favilli, S.
and Domenicucci, S.
and Russo, M. G.
and Sarubbi, B.
and Gelsomino, S.
and Colivicchi, F.
and Gulizia, M. M.},
title={Pregnancy and heart disease: the role of the Pregnancy Heart Team},
journal={GIORNALE ITALIANO DI CARDIOLOGIA},
year={AUG},
volume={23},
number={8},

pages={631-644},
keywords={Acquired heart disease; Congenital heart disease; Pregnancy Heart Team;
EUROPEAN-SOCIETY; CARDIAC-DISEASE; PERIPARTUM CARDIOMYOPATHY; SCIENTIFIC
STATEMENT; FAILURE ASSOCIATION; TASK-FORCE; HYPERTROPHIC CARDIOMYOPATHY;
VENOUS THROMBOEMBOLISM; HYPERTENSIVE DISORDERS; CARDIOVASCULAR-DISEASE},
abstract={A significant risk of maternal and fetal morbidity and mortality has been shown to be
associated with congenital heart disease or heart disease occurring during pregnancy. Given the
increasing number of patients with corrected congenital heart disease who reach fertile age and the
more and more common advanced maternal age associated with preexisting or intercurrent
comorbidities, a higher incidence of cardiac complications in pregnancy has been reported in the last
decades. Improvement in maternal and neonatal outcomes is influenced by a multidisciplinary
strategy. The purpose of this review is to assess the role of the Pregnancy Heart Team which should
ensure careful pre-pregnancy counseling, pregnancy monitoring, and delivery planning for both
congenital heart disease and other cardiac or metabolic disorders.},
note={Times Cited in Web of Science Core Collection: 1 Total Times Cited: 1 Cited Reference Count:
112},
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language={Italian}

@Article{MaenoDEC,
author={Maeno, Y.
and Himeno, W.
and Fujino, H.
and Sugahara, Y.
and Furui, J.
and Mizumoto, Y.
and Kato, H.},
title={Progression of congenital heart disease in the prenatal period},
journal={PEDIATRICS INTERNATIONAL},
year={DEC},
volume={41},
number={6},
pages={709-715},
keywords={congenital heart defect; development; fetal arrhythmia; fetal hydrops; prenatal
echocardiography; OUTFLOW TRACT OBSTRUCTION; IN-UTERO; FETAL ECHOCARDIOGRAPHY;
PREMATURE CLOSURE; PULMONARY ATRESIA; DUCTUS-ARTERIOSUS; AORTIC-STENOSIS;
FORAMEN OVALE; DIAGNOSIS; FETUS},
abstract={Background: Prenatal echocardiography has shown evidence of prenatal development of
congenital heart disease. Prenatal cardiac anatomy, chamber size and function change during
gestation, so that the appearance of cardiac structure in abnormal hearts may be different from that
which is usually seen postnatally. Methods: Published prenatal echocardiographic studies were
reviewed and in utero development of congenital heart disease from midtrimester to the early
postnatal period is discussed. Results: The growth of the great vessels and ventricles is reduced in
fetuses with ventricular outflow obstruction. Valve regurgitation may progress. The foramen ovale and
ductus arteriosus have been reported to become restrictive in utero in several settings. Pulmonary
vascular obstructive changes may progress prenatally. Fetal arrhythmia (both bradycardia and
tachycardia) may develop in utero. Development of congestive heart failure is a very important issue
during follow up of fetuses with significant cardiac or extra-cardiac problems. Some may progress to
fetal hydrops and prognosis of the affected fetuses is usually very poor. Conclusions: Correct
knowledge of possible development is important for accurate prenatal diagnosis. Information on

prenatal progression of the cardiac anomaly is also important to make plans for follow up and perinatal management, to predict outcomes and to counsel family. Furthermore, the benefits of prenatal treatment instead of postnatal treatment should be assessed by the accurate prediction of the progression of the cardiac problem in utero. Further extensive studies using a large number of cases is required to predict progression accurately. In addition, further studies for elucidating the mechanisms of progression is important to provide better outcomes for fetuses with various congenital heart diseases.},

note={Times Cited in Web of Science Core Collection: 11 Total Times Cited: 12 Cited Reference Count: 33},

issn={1328-8067},

doi={10.1046/j.1442-200x.1999.01153.x},

url={https://doi.org/10.1046/j.1442-200x.1999.01153.x},

language={English}

@Article{Ruan2021,

author={Ruan, Y. P.

and Liu, X. Y.

and Zhu, H. G.

and Lu, Y. J.

and Liu, X. W.

and Han, J. C.

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and Ran, S. Z.

and Chen, J. L.

and Yu, Q.

and Xu, Y.

and Xia, H. M.

and He, Y. H.},

title={Noninherited Factors in Fetal Congenital Heart Diseases Based on Bayesian Network: A Large Multicenter Study},

journal={CONGENITAL HEART DISEASE},

year={2021},

volume={16},

number={6},

pages={529-549},

keywords={Congenital heart diseases; bayesian network; risk ratio; factor; SCIENTIFIC STATEMENT; RISK-FACTORS; DEFECTS; ASSOCIATION; KNOWLEDGE; SMOKING; TOBACCO},

abstract={Background: Current studies have confirmed that fetal congenital heart diseases (CHDs) are caused by various factors. However, the quantitative risk of CHD is not clear given the combined effects of multiple factors. Objective: This cross-sectional study aimed to detect associated factors of fetal CHD using a Bayesian network in a large sample and quantitatively analyze relative risk ratios (RRs). Methods: Pregnant women who underwent fetal echocardiography (N = 16,086 including 3,312 with CHD fetuses) were analyzed. Twenty-six maternal and fetal factors were obtained. A Bayesian network is constructed based on all variables through structural learning and parameter learning methods to find the environmental factors that directly and indirectly associated with outcome, and the probability of fetal CHD in the two groups is predicted through a junction tree reasoning algorithm, so

as to obtain RR for fetal CHD under different exposure factor combinations. Taking into account the effect of gestational week on the accuracy of model prediction, we conducted sensitivity analysis on gestational week groups. Results: The single-factor analysis showed that the RRs for the numbers of births, spontaneous abortions, and parental smoking were 1.50, 1.38, and 1.11 ($P < 0.001$), respectively. The risk gradually increased with the synergistic effect of ranging from one to more environmental factors above. The risk was higher among subjects with five synergistic factors, including the number of births, upper respiratory tract infection during early pregnancy, anemia, and mental stress as well as a history of spontaneous abortions or parental smoking, than in those with less than 5 factors ($RR = 2.62$ or 2.28 , $P < 0.001$). This result was consistent across the participants grouped by GWs. Conclusion: We identified six factors that were directly associated with fetal CHD. A higher number of these factors led to a higher risk of CHD. These findings suggest that it is important to strengthen healthcare and prenatal counseling for women with these factors.},
note={Times Cited in Web of Science Core Collection: 0 Total Times Cited: 0 Cited Reference Count: 30},
issn={1747-079X},
language={English}

@Article{SonJAN,
author={Son, S. L.
and Hosek, L. L.
and Stein, M. C.
and Allshouse, A. A.
and Catino, A. B.
and Hoskoppal, A. K.
and Cox, D. A.
and Whitehead, K. J.
and Lindsay, I. M.
and Esplin, S.
and Metz, T. D.},
title={Association between pregnancy and long-term cardiac outcomes in individuals with congenital heart disease},
journal={AMERICAN JOURNAL OF OBSTETRICS AND GYNECOLOGY},
year={JAN},
volume={226},
number={1},
keywords={adult congenital heart disease; aortopathy; maternal aortopathy; maternal congenital heart disease; maternal heart disease; pregnancy with congenital heart disease; WOMEN},
abstract={BACKGROUND: As early life interventions for congenital heart disease improve, more patients are living to adulthood and are considering pregnancy. Scoring and classification systems predict the maternal cardiovascular risk of pregnancy in the context of congenital heart disease, but these scoring systems do not assess the potential subsequent risks following pregnancy. Data on the long-term cardiac outcomes after pregnancy are unknown for most lesion types. This limits the ability of healthcare practitioners to thoroughly counsel patients who are considering pregnancy in the setting of congenital heart disease. OBJECTIVE: We aimed to evaluate the association between pregnancy and the subsequent long-term cardiovascular health of individuals with congenital heart disease. STUDY DESIGN: This was a retrospective longitudinal cohort study of individuals identifying as female who were receiving care in two adult congenital heart disease centers from 2014 to 2019. Patient data were abstracted longitudinally from a patient age of 15 years (or from the time of entry into the healthcare system) to the conclusion of the study, death, or exit from the healthcare system. The primary endpoint, a composite adverse cardiac outcome (death, stroke, heart failure, unanticipated

cardiac surgery, or a requirement for a catheterized procedure), was compared between parous (at least one pregnancy >20 weeks' gestation) and nulliparous individuals. By accounting for differences in the follow-up, the effect of pregnancy was estimated based on the time to the composite adverse outcome in a proportional hazards regression model adjusted for the World Health Organization class, baseline cardiac medications, and number of previous sternotomies. Participants were also categorized according to their lesion type, including septal defects (ventricular septal defects, atrial septal defects, atrioventricular septal defects, or atrioventricular canal defects), right-sided valvular lesions, left-sided valvular lesions, complex cardiac anomalies, and aortopathies, to evaluate if there is a differential effect of pregnancy on the primary outcome when adjusting for lesion type in a sensitivity analysis. RESULTS: Overall, 711 individuals were eligible for inclusion; 209 were parous and 502 nulliparous. People were classified according to the World Health Organization classification system with 86 (12.3%) being classified as class I, 76 (10.9%) being classified as class II, 272 (38.9%) being classified as class II to III, 155 (22.1%) being classified as class III, and 26 (3.7%) being classified as class IV. Aortic stenosis, bicuspid aortic valve, dilated ascending aorta or aortic root, aortic regurgitation, and pulmonary insufficiency were more common in parous individuals, whereas dextro-transposition of the great arteries, Turner syndrome, hypoplastic right heart, left superior vena cava, and other cardiac diagnoses were more common in nulliparous individuals. In multivariable modeling, pregnancy was associated with the composite adverse cardiac outcome (36.4% vs 26.1%; hazard ratio, 1.83; 95% confidence interval, 1.25-2.66). Parous individuals were more likely to have unanticipated cardiac surgery (28.2% vs 18.1%; P=.003). No other individual components of the primary outcome were statistically different between parous and nulliparous individuals in cross-sectional comparisons. The association between pregnancy and the primary outcome was similar in a sensitivity analysis that adjusted for cardiac lesion type (hazard ratio, 1.61; 95% confidence interval, 1.10-2.36). CONCLUSION: Among individuals with congenital heart disease, pregnancy was associated with an increase in subsequent long-term adverse cardiac outcomes. These data may inform counseling of individuals with congenital heart disease who are considering pregnancy.},
note={124.e1-8},
note={Times Cited in Web of Science Core Collection: 1 Total Times Cited: 1 Cited Reference Count: 16},
issn={0002-9378},
doi={10.1016/j.ajog.2021.07.015},
url={https://doi.org/10.1016/j.ajog.2021.07.015},
language={English}

@Article{Bravo-valenzuelaJAN-FEB,
author={Bravo-valenzuela, N. J.
and Peixoto, A. B.
and Araujo, E.},
title={Prenatal diagnosis of congenital heart disease: A review of current knowledge},
journal={INDIAN HEART JOURNAL},
year={JAN},
month={FEB},
volume={70},
number={1},
pages={150-164},
keywords={Prenatal diagnosis; Ultrasound imaging; Echocardiography; Congenital heart disease; BIRTH PREVALENCE; TRICUSPID-VALVE; GREAT-ARTERIES; OUTFLOW TRACTS; SEPTAL-DEFECTS; Z-SCORES; FETAL; FETUS; ULTRASOUND; ANOMALIES},

abstract={This article reviews important features to improve the diagnosis of congenital heart disease (CHD) by applying ultrasound in prenatal cardiac screening. As low and high-risk pregnancies for CHD are subject to routine obstetric ultrasound, the diagnosis of structural heart defects represents a challenge that involves a team of specialists and subspecialists on fetal ultrasonography. In this review, the images highlight normal anatomy of the heart as well as pathologic cases consistent with cardiac malposition and isomerism, septal defects, pulmonary stenosis/atresia, aortic malformations, hypoplastic left ventricle, conotruncal anomalies, tricuspid dysplasia, and Ebstein's anomaly, and univentricular heart, among other congenital cardiovascular defects. Anatomical details of most CHD in fetuses were provided by two-dimensional (2D) ultrasound with higher quality imaging, enhancing diagnostic accuracy in a variety of CHD. Moreover, the accuracy of the cardiac defects in obstetrics ultrasound improves the outcome of most CHD, providing planned delivery, aided genetic counseling, and perinatal management. (C) 2017 Cardiological Society of India. Published by Elsevier B.V.},
note={Times Cited in Web of Science Core Collection: 28 Total Times Cited: 34 Cited Reference Count: 59},
issn={0019-4832},
doi={10.1016/j.ihj.2017.12.005},
url={https://doi.org/10.1016/j.ihj.2017.12.005},
language={English}

@Article{FoellerJUN,
author={Foeller, M. E.
and Foeller, T. M.
and Druzin, M.},
title={Maternal Congenital Heart Disease in Pregnancy},
journal={OBSTETRICS AND GYNECOLOGY CLINICS OF NORTH AMERICA},
year={JUN},
volume={45},
number={2},
pages={267-+},
keywords={Congenital heart disease; Pregnancy; Tetralogy of Fallot; Transposition of great arteries; Pulmonary hypertension; EISENMENGER SYNDROME; CARDIAC OUTCOMES; SEPTAL-DEFECT; WOMEN; ADULT; MANAGEMENT; REPAIR; RISK; HYPERTENSION; COARCTATION},
abstract={Congenital heart disease comprises most maternal cardiac diseases in pregnancy and is an important cause of maternal, fetal, and neonatal morbidity and mortality worldwide. Pregnancy is often considered a high risk state for individuals with structural heart disease as a consequence of a limited ability to adapt to the major hemodynamic changes associated with pregnancy. Preconception counseling and evaluation are of utmost importance, as pregnancy is contraindicated in certain cardiac conditions. Pregnancy can be safely accomplished in most individuals with careful risk assessment before conception and multidisciplinary care throughout pregnancy and the postpartum period.},
note={Times Cited in Web of Science Core Collection: 4 Total Times Cited: 6 Cited Reference Count: 58},
issn={0889-8545},
doi={10.1016/j.ogc.2018.01.011},
url={https://doi.org/10.1016/j.ogc.2018.01.011},
language={English}

@Article{KovacevicAUGb,
author={Kovacevic, A.

and Bar, S.
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and van der Locht, T.
and Motlagh, A. M.
and Ostermayer, E.
and Oberhoffer-Fritz, R.
and Ewert, P.
and Gorenflo, M.
and Wacker-Gussmann, A.},

title={Fetal Cardiac Services during the COVID-19 Pandemic: How Does It Affect Parental Counseling?},

journal={JOURNAL OF CLINICAL MEDICINE},

year={AUG},

volume={10},

number={15},

keywords={COVID-19 pandemic; fetal congenital heart disease; prenatal diagnosis; parental counseling; MATERNAL STRESS; DISEASE; PREGNANCY; DIAGNOSIS; DEPRESSION},

abstract={The COVID-19 pandemic impacts health care providers in multiple ways, even specialties that do not seem to be affected primarily, such as fetal cardiac services. We aimed to assess the effects on parental counseling for fetal congenital heart disease (CHD). In this multicenter study, we used a validated questionnaire. Parents were recruited from four national tertiary medical care centers (n = 226); n = 169 had been counseled before and n = 57 during the pandemic. Overall counseling success including its dimensions did not differ between the two groups (p = n.s.). However, by applying the sorrow scale, we could demonstrate that parents counseled during the pandemic were significantly more concerned (p = 0.025) and unsure (p = 0.044) about their child's diagnosis, therapy and outcome. Furthermore, parents expressed a significantly increased need for written and/or online information on fetal heart disease (p = 0.034). Other modifiers did not affect counseling success (p = n.s.). We demonstrate that the COVID-19 pandemic impacts effectiveness of parental counseling for fetal CHD, possibly by altering parental perceptions. This needs to be taken into consideration when counseling. Implementing alternative and innovative approaches (e.g., online conference or virtual reality tools) may aid in facilitating high-quality services in critical times such as in the present pandemic.},

note={3423},

note={Times Cited in Web of Science Core Collection: 1 Total Times Cited: 1 Cited Reference Count: 27},

issn={2077-0383},

doi={10.3390/jcm10153423},

url={https://doi.org/10.3390/jcm10153423},

language={English}

@Article{SchmaltzJAN,

author={Schmaltz, A. A.

and Neudorf, U.

and Winkler, U. H.},

title={Outcome of pregnancy in women with congenital heart disease},

journal={CARDIOLOGY IN THE YOUNG},

year={JAN},

volume={9},

number={1},

pages={88-96},
keywords={pregnancy; congenital heart disease; maternal risk; fetal risk; anticoagulation; genetic risk; BALLOON AORTIC VALVULOPLASTY; FAMILY-PLANNING REQUIREMENTS; FONTAN OPERATION; FOLLOW-UP; ANTICOAGULANT-THERAPY; EISENMENGENERS-SYNDROME; VALVULAR PROSTHESES; CARDIAC-OUTPUT; MATERNAL HEART; VALVES},
abstract={Improvements in diagnosis and surgical technique for correction have led to an increasing number of women with congenital heart disease reaching the child-bearing age. Pregnancy places considerable strain on the heart and circulation and necessitates marked cardiorespiratory adaptation. Today, with the exception of the Eisenmenger syndrome, there is no increased mortality associated with pregnancy in congenital heart disease. In contrast, there is still considerable morbidity, due to congestive heart failure, thromboembolic complications and disturbances of rhythm. Fetal outcome is complicated by a high rate of spontaneous abortions (20-25{\%}), retardation of fetal growth, and premature delivery (almost 100{\%} in cyanotic mothers), Based on an extensive review of the literature, we discuss the specific risks in pregnancy depending on the hemodynamic situations produced by different heart defects. We also discuss the risks and advantages of different regimens for anticoagulation. Counselling concerning contraception is frequently inadequate. The most important problems are thromboembolic complications with the use of hormonal contraception, and hyper- and dysmenorrhea in those using intrauterine devices. Finally, the genetic risks must be considered, differentiating between single gene defects and the sex of the parents suffering from congenital heart diseases.},
note={Times Cited in Web of Science Core Collection: 9 Total Times Cited: 10 Cited Reference Count: 68},
issn={1047-9511},
doi={10.1017/S1047951100007496},
url={https://doi.org/10.1017/S1047951100007496},
language={English}

@Article{GoncalvesMAR,
author={Goncalves, L. F.
and Lindblade, C. L.
and Cornejo, P.
and Patel, M. C.
and McLaughlin, E. S.
and Bardo, D. M. E.},
title={Contribution of fetal magnetic resonance imaging in fetuses with congenital heart disease},
journal={PEDIATRIC RADIOLOGY},
year={MAR},
volume={52},
number={3},
pages={513-526},
keywords={Anomalies; Brain; Congenital heart disease; Echocardiography; Fetus; Heart; Magnetic resonance imaging; Prenatal ultrasound; Pulmonary lymphangiectasia; BRAIN ABNORMALITIES; PULMONARY LYMPHANGIECTASIS; PRENATAL-DIAGNOSIS; IN-UTERO; ULTRASOUND; DOPPLER; MRI},
abstract={Background Increasing evidence supports an association among congenital heart disease (CHD), structural brain lesions on neuroimaging, and increased risk of neurodevelopmental delay and other structural anomalies. Fetal MRI has been found to be effective in demonstrating fetal structural and developmental abnormalities. Objective To determine the contribution of fetal MRI to identifying cardiovascular and non-cardiovascular anomalies in fetuses with CHD compared to prenatal US and fetal echocardiography. Materials and methods We performed a retrospective study of fetuses with

CHD identified by fetal echocardiography. Exams were performed on 1.5-tesla (T) or 3-T magnets using a balanced turbo field echo sequence triggered by an external electrocardiogram simulator with a fixed heart rate of 140 beats per minute (bpm). Fetal echocardiography was performed by pediatric cardiologists and detailed obstetrical US by maternal-fetal medicine specialists prior to referral to MRI. We compared the sensitivity of fetal MRI and fetal echocardiography for the diagnosis of cardiovascular anomalies, as well as the sensitivity of fetal MRI and referral US for the diagnosis of non-cardiac anomalies. We performed statistical analysis using the McNemar test. Results We identified 121 anomalies in 31 fetuses. Of these, 73 (60.3%) were cardiovascular and 48 (39.7%) involved other organ systems. Fetal echocardiography was more sensitive for diagnosing cardiovascular anomalies compared to fetal MRI, but the difference was not statistically significant (85.9%, 95% confidence interval [CI] 77.8-94.0% vs. 77.5%, 95% CI 67.7-87.2%, respectively; McNemar test 2.29; P=0.13). The sensitivity of fetal MRI was higher for diagnosing extracardiac anomalies when compared to referral US (84.1%, 95% CI 73.3-94.9% vs. 31.8%, 95% CI 18.1-45.6%, respectively; McNemar test 12.9; P<0.001). The additional information provided by fetal MRI changed prognosis, counseling or management for 10/31 fetuses (32.2%), all in the group of 19 fetuses with anomalies in other organs and systems besides CHD. Conclusion Fetal MRI performed in a population of fetuses with CHD provided additional information that altered prognosis, counseling or management in approximately one-third of the fetuses, mainly by identifying previously unknown anomalies in other organs and systems.},

note={NOV 2021},

note={Times Cited in Web of Science Core Collection: 0 Total Times Cited: 0 Cited Reference Count: 37},

issn={0301-0449},

doi={10.1007/s00247-021-05234-1},

url={https://doi.org/10.1007/s00247-021-05234-1},

language={English}

@Article{ref36,

author={Ntiloudi, D.

and Zegkos, T.

and Koutsakis, A.

and Giannakoulas, G.

and Karvounis, H.},

title={Pregnancy in Patients With Congenital Heart Disease A Contemporary Challenge},

journal={CARDIOLOGY IN REVIEW},

keywords={pregnancy; congenital heart disease; cardiac complications; obstetric complications; fetal complications; EUROPEAN-SOCIETY; FONTAN CIRCULATION; RISK-ASSESSMENT; WOMEN; OUTCOMES; REGISTRY; COMPLICATIONS; ROPAC; HYPERTENSION; POPULATION},

abstract={The majority of female patients with congenital heart disease (CHD) survives into childbearing age and require evidence-based counseling regarding pregnancy options. Even though most of them will have an uneventful pregnancy, they may be at high risk of cardiac, obstetric, and fetal complications. Predictive factors for these complications have been previously identified in numerous studies and with the use of specific scores [CARDiac disease in PREGnancy,

Zwangerschap bij Aangeboren HA},

language={English}

APÊNDICE G - RESULTADOS DE BUSCA NO PSYCINFO

@Article{McKnight-Eily2017,
author={McKnight-Eily, Lela R.
and Henley, S. Jane
and Green, Patricia P.
and Odom, Erika C.
and Hungerford, Daniel W.},
title={Alcohol screening and brief intervention: A potential role in cancer prevention for young adults.},
journal={American Journal of Preventive Medicine},
year={2017},
publisher={Elsevier Science},
address={McKnight-Eily, Lela R.: 1600 Clifton Road, NE, E86, Atlanta, GA, US, 30333,
dvn1@cdc.gov},
volume={53},
pages={S55-S62},
keywords={*Health Care Services; *Primary Health Care; Cancer Screening; Counseling},
abstract={Excessive or risky alcohol use is a preventable cause of significant morbidity and mortality in the U.S. and worldwide. Alcohol use is a common preventable cancer risk factor among young adults; it is associated with increased risk of developing at least six types of cancer. Alcohol consumed during early adulthood may pose a higher risk of female breast cancer than alcohol consumed later in life. Reducing alcohol use may help prevent cancer. Alcohol misuse screening and brief counseling or intervention (also called alcohol screening and brief intervention among other designations) is known to reduce excessive alcohol use, and the U.S. Preventive Services Task Force recommends that it be implemented for all adults aged ≥ 18 years in primary healthcare settings. Because the prevalence of excessive alcohol use, particularly binge drinking, peaks among young adults, this time of life may present a unique window of opportunity to talk about the cancer risk associated with alcohol use and how to reduce that risk by reducing excessive drinking or misuse. This article briefly describes alcohol screening and brief intervention, including the Centers for Disease Control and Prevention's recommended approach, and suggests a role for it in the context of cancer prevention. The article also briefly discusses how the Centers for Disease Control and Prevention is working to make alcohol screening and brief intervention a routine element of health care in all primary care settings to identify and help young adults who drink too much. (PsycINFO Database Record (c) 2019 APA, all rights reserved)},
doi={10.1016/j.amepre.2017.04.021},
url={https://doi.org/10.1016/j.amepre.2017.04.021}}

@Article{Honicky2020,
author={Honicky, M.
and Cardoso, S. M.
and Lima, L. R. A.
and Ozcariz, S. G. I.
and Vieira, F. G. K.
and Carlos Back, I.
and Moreno, Y. M. F.},
title={Added sugar and trans fatty acid intake and sedentary behavior were associated with excess total-body and central adiposity in children and adolescents with congenital heart disease.},
journal={Pediatric Obesity},

year={2020},
publisher={Wiley-Blackwell Publishing Ltd.},
address={Moreno, Y. M. F.: Postgraduate Program in Nutrition, Federal University of Santa Catarina, Health Sciences Center, University Campus, Trindade, SC, Florianopolis, Brazil, CEP 88040-900, yara.moreno@ufsc.br},
volume={15},
keywords={*Body Fat; *Fatty Acids; *Heart Disorders; *Obesity; *Sugars; Food Intake; Lifestyle; Pediatrics; Physique; Sedentary Behavior},
abstract={Background: Over the past three decades, the prevalence rate of overweight and obesity has increased in survivors with congenital heart disease, and little is known about the body composition and its association with clinical characteristics and lifestyle factors. Objectives: To evaluate excess total-body adiposity and central adiposity and, to describe associated factors. Methods: Cross-sectional study with children and adolescents who underwent procedure to treat congenital heart disease, from January to July 2017. Sociodemographic and clinical characteristics, and lifestyle factors (dietary intake, physical activity, and sedentary behavior) were assessed. Adiposity was assessed using air-displacement plethysmography and waist circumference. Factors associated with excess total-body adiposity and central adiposity were analyzed using logistic regression models. Results: Of 232 patients, 22.4{\%} were identified with excess total-body adiposity and 24.6{\%} with central adiposity. Significant factors positively associated with excess total-body adiposity were intake of added sugar and trans fatty acids, adjusted for confounding factors. Similarly, lifestyle factors were positively associated with central adiposity: intake of added sugar and trans fatty acids, sedentary behavior, and family history of obesity. Conclusions: Lifestyle factors were associated with excess total-body adiposity and central adiposity. Assessment of body composition and healthy-lifestyle counseling into outpatient care may be the key point to prevent obesity in children and adolescents with congenital heart disease. (PsycInfo Database Record (c) 2020 APA, all rights reserved)},
doi={10.1111/ijpo.12623},
url={https://doi.org/10.1111/ijpo.12623}

@Article{Ison2021,
author={Ison, Hannah E.
and Griffin, Emily L.
and Parrott, Ashley
and Shikany, Amy R.
and Meyers, Lindsay
and Thomas, Matthew J.
and Syverson, Erin
and Demo, Erin M.
and Fitzgerald, Kristi K.
and Fitzgerald-Butt, Sara
and Ziegler, Katie L.
and Schartman, Allison F.
and Stone, Kristyne M.
and Helm, Benjamin M.},
title={Genetic counseling for congenital heart disease -- practice resource of the national society of genetic counselors.},
journal={Journal of Genetic Counseling},
year={2021},
publisher={Wiley-Blackwell Publishing Ltd.},
address={United Kingdom},

pages={No Pagination Specified-No Pagination Specified},
abstract={Congenital heart disease (CHD) is an indication which spans multiple specialties across various genetic counseling practices. This practice resource aims to provide guidance on key considerations when approaching counseling for this particular indication while recognizing the rapidly changing landscape of knowledge within this domain. This resource was developed with consensus from a diverse group of certified genetic counselors utilizing literature relevant for CHD genetic counseling practice and is aimed at supporting genetic counselors who encounter this indication in their practice both pre- and postnatally. (PsycInfo Database Record (c) 2021 APA, all rights reserved)},
doi={10.1002/jgc4.1498},
url={https://doi.org/10.1002/jgc4.1498}

@Article{Pagé2012,
author={Pagé, M. Gabrielle
and Kovacs, Adrienne H.
and Irvine, Jane},
title={How do psychosocial challenges associated with living with congenital heart disease translate into treatment interests and preferences? A qualitative approach.},
journal={Psychology {&} Health},
year={2012},
publisher={Taylor {&} Francis},
address={Kovacs, Adrienne H.: Toronto Congenital Cardiac Centre for Adults, Peter Munk Cardiac Centre, University Health Network, Toronto, ON, Canada, adrienne.kovacs@uhn.ca},
volume={27},
pages={1260-1270},
keywords={*Congenital Disorders; *Heart Disorders; *Psychosocial Factors; *Treatment; Counseling},
abstract={Objectives: There is an increasing amount of research being conducted regarding the psychosocial challenges associated with living with congenital heart disease (CHD), however little is known about how these challenges influence the type of psychosocial services patients want. This study investigated (1) the type of services patients want; (2) how they want to access these services; and (3) why they want these services. Methods: Three focus groups with adults with CHD (total of 14 participants aged 19–67) were conducted and thematic analysis was used to identify emerging themes. Results: Participants described wanting to access specific psychosocial services in three broad categories (counseling, connecting with other adults with CHD and psycho-education) and in three main formats (individual/group therapy, mentorship programmes and patient conferences). Reasons for wanting these services were grouped under two overarching themes, namely intrapersonal factors and interpersonal challenges. Conclusions: Psychosocial challenges are part of the everyday lives of adults with CHD, yet they are rarely addressed as part of routine medical care. Patients themselves have clear opinions regarding the psychological services most appropriate to target their experiences of living with CHD. (PsycINFO Database Record (c) 2016 APA, all rights reserved)},
doi={10.1080/08870446.2012.667099},
url={https://doi.org/10.1080/08870446.2012.667099}}

@Article{Gendler2021,
author={Gendler, Yulia
and Birk, Einat
and Tabak, Nili}

and Koton, Silvia},
title={Factors that influence parents' decision-making regarding termination of pregnancy after prenatal diagnosis of fetal congenital heart disease.},
journal={Journal of Obstetric, Gynecologic, {\&} Neonatal Nursing: Clinical Scholarship for the Care of Women, Childbearing Families, {\&} Newborns},
year={2021},
publisher={Elsevier Science},
address={Gendler, Yulia: Department of Nursing, Ariel University, 65 Ramat-haGolan Street, Ariel, Israel, yulia.gendler@gmail.com},
volume={50},
pages={475-484},
keywords={*Decision Making; *Fetus; *Heart Disorders; *Pregnancy; *Prenatal Diagnosis; Couples; Pediatrics; Prospective Studies},
abstract={Objective: To explore factors that influence parents' decisions regarding the termination of pregnancy after the detection of fetal congenital heart disease (CHD). Design: A prospective descriptive study. Setting: The Institute of Pediatric Cardiology in the Schneider Children's Medical Center. Participants: One hundred twenty couples (240 participants) with fetuses prenatally diagnosed with CHD, which was defined as conditions requiring surgical treatment. Methods: We obtained data from a structured questionnaire for the pediatric cardiologist, the medical records, and structured self-report questionnaires for the participants. Results: Thirty-six of 120 couples (30{\%}) decided to terminate the pregnancy after a prenatal diagnosis of fetal CHD. The main factors associated with the decision to terminate were low gestational age (OR = 0.83 per week, 95{\%} confidence interval [CI] [0.75, 0.96]), severe cardiac malformation (OR = 2.23, 95{\%} CI [1.40, 3.53]), religious affiliation (OR = 10.0 for secular participants vs. others, 95{\%} CI [4.61, 22.46]), population group (OR = 2.96 for Jewish participants vs. others, 95{\%} CI [1.63, 11.3]), and education (OR = 1.34 per year of education, 95{\%} CI [1.15, 1.55]). Conclusions: Our findings describe the profiles of couples who decided to terminate their pregnancies after a prenatal diagnosis of fetal CHD. Early identification of the couple's decision can help health care providers provide adequate support, counseling, and guidance. Future research is needed to understand parents' needs for support through the process of decision-making, with attention to their religious and cultural values and contexts. (PsycInfo Database Record (c) 2022 APA, all rights reserved)},
doi={10.1016/j.jogn.2021.04.002},
url={https://doi.org/10.1016/j.jogn.2021.04.002}

@Article{Tabata2017,
author={Tabata, Hisae},
title={Care of preschoolers with congenital heart disease by kindergarten and nursery teachers in Japan.},
journal={Comprehensive Child and Adolescent Nursing},
year={2017},
publisher={Taylor {\&} Francis},
address={Tabata, Hisae: Department of Nursing, School of Health Sciences, Sapporo Medical University, Minami 1, Nishi 17, Chuo-ku, Hokkaido, Sapporo, Japan, 060-8556, hisaet@sapmed.ac.jp},
volume={40},
pages={144-156},
keywords={*Congenital Disorders; *Elementary School Teachers; *Heart Disorders; *Nursery Schools; *Preschool Students; Child Care},
abstract={The purpose of this study was to elucidate the involvement of kindergarten and nursery school teachers with young children with congenital heart disease. The study was designed as a qualitative descriptive study. Interviews of kindergarten and nursery school teachers with experience in

the care and education of young children with congenital heart disease were conducted, during which they described their experience. Verbatim transcripts of the interviews were prepared, and the content was categorized. The study participants were 11 kindergarten and nursery school teachers. Extracted from the content of the interviews of the study participants were 282 codes, 33 subcategories, 6 categories, and 2 major categories. In their responses, the teachers indicated that they had been "Providing care for the children while seeking ways to avoid special treatment in a group setting." In addition, they established a "Framework for school--parent cooperation in order to promptly accommodate the wishes of parents" of these children. The study showed that the kindergarten and nursery school teachers involved other pupils and monitored the condition of children with congenital heart disease to avoid special treatment of the children in the group setting. In addition, the teachers established a framework for cooperation between the school and parents. In the future, these findings will be used to create a nursing support model for the group life of young children with congenital heart disease. (PsycINFO Database Record (c) 2019 APA, all rights reserved)},
doi={10.1080/24694193.2017.1307472},
url={https://doi.org/10.1080/24694193.2017.1307472}

@Article{Freeze2016,
author={Freeze, Samantha L.
and Landis, Benjamin J.
and Ware, Stephanie M.
and Helm, Benjamin M.},
title={Bicuspid aortic valve: A review with recommendations for genetic counseling.},
journal={Journal of Genetic Counseling},
year={2016},
publisher={Springer},
address={Helm, Benjamin M.: Department of Medical {&} Molecular Genetics, Riley Hospital for Children at IU Health, Indiana University School of Medicine, 975 West Walnut Street, IB-130, Indianapolis, IN, US, 46202, bmhelm@iu.edu},
volume={25},
pages={1171-1178},
keywords={*Congenital Disorders; *Counseling; *Genetics; Health Care Services},
abstract={Bicuspid aortic valve (BAV) is the most common congenital heart defect and falls in the spectrum of left-sided heart defects, also known as left ventricular outflow tract obstructive (LVOTO) defects. BAV is often identified in otherwise healthy, asymptomatic individuals, but it is associated with serious long term health risks including progressive aortic valve disease (stenosis or regurgitation) and thoracic aortic aneurysm and dissection. BAV and other LVOTO defects have high heritability. Although recommendations for cardiac screening of BAV in at-risk relatives exist, there are no standard guidelines for providing genetic counseling to patients and families with BAV. This review describes current knowledge of BAV and associated aortopathy and provides guidance to genetic counselors involved in the care of patients and families with these malformations. The heritability of BAV and recommendations for screening are highlighted. While this review focuses specifically on BAV, the principles are applicable to counseling needs for other LVOTO defects. (PsycINFO Database Record (c) 2019 APA, all rights reserved)},
doi={10.1007/s10897-016-0002-6},
url={https://doi.org/10.1007/s10897-016-0002-6}

@Article{Menahem2005,
author={Menahem, Samuel

and Grimwade, James},
title={Pre-natal counselling--helping couples make decisions following the diagnosis of severe heart disease.},
journal={Early Human Development},
year={2005},
publisher={Elsevier Science},
address={Menahem, Samuel: Monash University Heart and Chest Research Institute, Department of Psychological Medicine, Monash University, Monash Medical Centre, 246 Clayton Road, Clayton, VIC, Australia, 3168, sam.menahem@rch.org.au},
volume={81},
pages={601-607},
keywords={*Counseling; *Couples; *Decision Making; *Heart Disorders; *Prenatal Diagnosis; Diagnosis; Parents},
abstract={The prenatal diagnosis of a major cardiac abnormality tends to precipitate a crisis for the affected parents. In a setting of grief and emotional distress, there is the challenge to provide meaningful information of the abnormality, its need for intervention and likely outcome, so as to enable the parents, if allowed the option, to come to a fully informed decision as to whether to continue with the pregnancy. This discussion paper reviews the difficulties encountered in counselling affected parents being mindful of the psychological constraints prevalent at the time. While an accurate and detailed diagnosis is important for the professionals, the information required by the parents needs to be simple and focussed on the questions raised by them as they relate to quality of life issues to be experienced by their yet unborn infant/child growing into an adult. (PsycINFO Database Record (c) 2017 APA, all rights reserved)},
doi={10.1016/j.earhumdev.2005.02.001},
url={https://doi.org/10.1016/j.earhumdev.2005.02.001}

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author={Derbent, M.
and Tokel, K.
and Saygili, A.
and Akkuzu, B.
and Oto, S.
and Co{\c{s}}kun, M.
and Balci, S.},
title={Middle and inner ear anomalies in a patient with charge association.},
journal={Genetic Counseling: Medical, Psychological, and Ethical Aspects},
year={2003},
publisher={Editions M{\e}decine et Hygi{\e}ne},
address={Balci, S.: Department of Clinical Genetics, Hacettepe University Faculty of Medicine, Sıhhiye, Ankara, Turkey, 06100},
volume={14},
pages={367-368},
keywords={*Congenital Disorders; *Ear Disorders; *Heart Disorders; *Labyrinth (Anatomy); Middle Ear},
abstract={Letter to the editor in response to the article "Middle And Inner Ear Anomalies In A Patient With Charge Association." The congenital anomalies are known as CHARGE association (CA) (coloboma of the eye [G], heart defects [H], choanal atresia [A], retardation of growth and/or development [R], genital hypoplasia or anomalies [G], and ear anomalies and/or deafness [E]. A 20-day-old female infant was referred to our hospital with congenital heart disease. Axial CT scans of both ears showed osseous atresia of the external auditory canals, dysplasia of the semicircular canals

bilaterally, and single-cavity vestibules. The auditory ossicles were malformed bilaterally, but both blood sample revealed normal karyotype. As our case demonstrates, CA patients may exhibit not only external and middle ear anomalies, but also inner ear defects such as semicircular canal dysplasia. Recently, authors reported three patients with CA who have neither coloboma nor choanal atresia, and have temporal bone anomalies including semicircular canal agenesis/hypoplasia. It is suggested that temporal bone anomalies should be regarded as a major diagnostic criteria for diagnosis of CA (1). (PsycINFO Database Record (c) 2016 APA, all rights reserved)},
issn={1015-8146(Print)}

@Article{Sigmon2019,
author={Sigmon, Eric R.
and Kelleman, Michael
and Susi, Apryl
and Nylund, Cade M.
and Oster, Matthew E.},
title={Congenital heart disease and autism: A case-control study.},
journal={Pediatrics},
year={2019},
publisher={American Academy of Pediatrics},
address={Oster, Matthew E.: Children's Healthcare of Atlanta, 2835 Brandywine Rd, Atlanta, GA, US, 30341, osterm@kidsheart.com},
volume={144},
keywords={*Autism Spectrum Disorders; *Comorbidity; *Congenital Disorders; *Heart Disorders; *Lesions; Military Families},
abstract={Objectives: There has long been an association between congenital heart disease (CHD) and general neurodevelopmental delays. However, the association between CHD and autism spectrum disorders (AuSDs) is less well understood. Using administrative data, we sought to determine the association between CHD and AuSD and identify specific CHD lesions with higher odds of developing AuSD. Methods: We performed a 1:3 case-control study of children enrolled in the US Military Health System from 2001 to 2013. Children with International Classification of Disease, Ninth Revision, Clinical Modification codes for AuSD were identified as cases and matched with controls on the basis of date of birth, sex, and enrollment time frame. Each child's records were reviewed for CHD lesions and associated procedures. Conditional logistic regression determined odds ratios (ORs) and 95% confidence intervals (CIs) for comparative associations. Results: There were 8760 cases with AuSD and 26 280 controls included in the study. After adjustment for genetic syndrome, maternal age, gestational diabetes, short gestation, newborn epilepsy, birth asphyxia, and low birth weight, there were increased odds of AuSD in patients with CHD (OR 1.32; 95% CI 1.10--1.59). Specific lesions with significant OR included atrial septal defects (n = 82; OR 1.72; 95% CI 1.07--2.74) and ventricular septal defects (n = 193; OR 1.65; 95% CI 1.21--2.25). Conclusion: Children with CHD have increased odds of developing AuSD. Specific lesions associated with increased risk include atrial septal defects and ventricular septal defects. These findings will be useful for counseling parents of children with CHD. (PsycInfo Database Record (c) 2020 APA, all rights reserved)},
doi={10.1542/peds.2018-4114},
url={https://doi.org/10.1542/peds.2018-4114}}

@Article{Somers2014,
author={Somers, Allyson E.
and Ware, Stephanie M.

and Collins, Kathleen
and Jefferies, John L.
and He, Hua
and Miller, Erin M.},
title={Provision of cardiovascular genetic counseling services: Current practice and future directions.},
journal={Journal of Genetic Counseling},
year={2014},
publisher={Springer},
address={Miller, Erin M.: Cincinnati Children's Hospital Medical Center, 3333 Burnet Avenue, MLC
7020, Cincinnati, OH, US, 45229, Erin.Miller@cchmc.org},
volume={23},
pages={976-983},
keywords={*Genetic Counseling; *Heart Disorders; *Training; Cardiovascular Disorders; Continuing
Education; Genetics},
abstract={Cardiovascular genetic counseling has emerged as a specialty critical to the care of patients
with heritable cardiovascular disease. Current strategies to meet the growing demand are not clear.
We sought to characterize practice patterns of cardiac genetic counseling by developing a novel
survey distributed to the National Society of Genetic Counselors (NSGC) Listserv to assess clinical
practice, cardiovascular training, and education. Descriptive statistics were used to summarize clinical
practice; Fisher's exact test and the Cochran-Armitage trend test were used to compare the practice of
cardiovascular genetic counselors (CVGCs) to those who did not identify cardiology as a specialty
(non-CVGCs). A total of 153 individuals completed the survey. Of the 105 participants who reported
seeing a cardiac genetics patient, 42 (40 { \% }) identified themselves as a CVGC. The most common
conditions for which genetic counseling was provided were hypertrophic cardiomyopathy (HCM) (71
{ \% }) of participants), dilated cardiomyopathy (DCM) (61 { \% }), long QT syndrome (LQTS) (56 { \% }
, and genetic syndromes with cardiovascular disease (55 { \% }). CVGCs were significantly more
confident than non-CVGCs in providing genetic counseling for seven cardiovascular diseases (2.3
{ \texttimes } 10-6 ≤ p ≤ 0.021). Eighty-six percent of genetic counselors sought additional education
related to cardiovascular genetics and listed online courses as the most desirable method of learning.
These data suggest a growing interest in cardiovascular genetic counseling and need for additional
training resources among the NSGC membership. (PsycINFO Database Record (c) 2019 APA, all
rights reserved)},
doi={10.1007/s10897-014-9719-2},
url={https://doi.org/10.1007/s10897-014-9719-2}

@Inbook{Campioni2015,
author={Campioni, Giovanna
and Callus, Edward
and Quadri, Emilia},
title={The role of associations in congenital heart disease: Peer counseling and advocacy.},
bookTitle={Clinical psychology and congenital heart disease: Lifelong psychological aspects and
interventions.},
year={2015},
publisher={Springer-Verlag Publishing},
address={Campioni, Giovanna: Associazione Italiana dei Cardiopatici Congeniti Adulti (AICCA),
Italian GUCH Association, Via G. Pascoli, 37 San Donato Milanese, Milan, Italy, 20097,
giocampioni@libero.it},
pages={189-199},
keywords={*Advocacy; *Congenital Disorders; *Heart Disorders; Peer Counseling},

abstract={In recent years, the number of voluntary, nonprofit associations formed by patients, created around the issues of a given disease, has increased. The formation of these associations normally arises from the need to meet, more broadly, some services provided by the national health system and to carry out initiatives for the protection and health and social care. In many cases, the complexity of the situation has led to a progressive organization of individual associations getting together and forming federations to give more strength to their message---both nationally and internationally---without losing their individual characteristics. In this way, requests for good quality care are becoming stronger and equally available nationwide, as well the dissemination of information about diseases and possibly their prevention or early detection. This also creates favorable conditions for investment in scientific research to improve the knowledge about diseases and to protect the patients and their families. In fact, the demand by citizens and patients to play an active role in the choices on health, both in the doctor-patient relationship---that is, increasingly moving away from a paternalistic vision of the role of the physician and it is becoming a relationship in which choices are shared---and also in health policy and research, has been growing, for several years. (PsycInfo Database Record (c) 2020 APA, all rights reserved)},
isbn={978-88-470-5698-5 (Hardcover); 978-88-470-5699-2 (Digital (undefined format))},
doi={10.1007/978-88-470-5699-2_13},
url={https://doi.org/10.1007/978-88-470-5699-2_13}

@Article{Kriss1999,
author={Kriss, Vesna Martich},
title={Down syndrome: Imaging of multiorgan involvement.},
journal={Clinical Pediatrics},
year={1999},
publisher={Westminster Publications},
address={US},
volume={38},
pages={441-449},
keywords={*Down's Syndrome; Tomography},
abstract={Down syndrome (trisomy 21) has many manifestations that affect multiple organ systems, and the authors describe the wide array of imaging findings. Common cardiovascular and gastrointestinal entities are congenital heart disease (atrioventricular canal), bowel atresias (duodenal and anal), and Hirschsprungs; disease. Children with Down syndrome have an 18--20 fold increased incidence of leukemia. Pulmonary hypoplasia, lung cysts, and pig bronchus (origin of the right upper lobe bronchus from the trachea) have been described. Neurologic findings include mineralizing vasculopathy of the basal ganglia, Moyamoya disease, and cerebellar/vermian hypoplasia. Musculoskeletal manifestations are numerous and include eleven ribs, hypersegmented sternum, abnormal pelvis, joint laxity/dislocations, and DDH (developmental dysplasia of the hip). Of special importance is the "triple jeopardy" of the upper cervical spine (atlanoaxial subluxation, hypoplastic posterior arch of C1, and atlantooccipital instability) and the resulting controversial cervical spine radiographic screening of children with Down syndrome. Knowledge of the many anomalies associated with Down syndrome can aid the clinician, not only in diagnosing abnormalities in these patients, but also in counseling families for potential problems that can occur in these children. (PsycINFO Database Record (c) 2016 APA, all rights reserved)},
issn={0009-9228(Print)},
doi={10.1177/000992289903800801},
url={https://doi.org/10.1177/000992289903800801}}

@Article{Piper1981,
author={Piper, Martha C.
and Lippman-Hand, Abby},
title={The convenience sample as a source of data in the study of Down syndrome.},
journal={Journal of Mental Deficiency Research},
year={1981},
publisher={Blackwell Publishing},
address={United Kingdom},
volume={25},
pages={217-223},
keywords={*Data Collection; *Down's Syndrome; *Sampling (Experimental); Statistical Samples},
abstract={Compared a total population of children with Down syndrome, live-born in Montreal in 1975 and 1976 (N=88) with 2 convenience samples drawn from this population: (a) cases known to a hospital genetic counseling center, and (b) cases participating in an early intervention program. Complete information on maternal age and parity, birth weight and sex of child, presence or absence of congenital heart disease, and residential placement was obtained for 83 members of the cohort. Analysis revealed that the cases in sample (a) were more likely to have younger mothers caring for them at home than the remaining cases in the Down's syndrome population; in sample (b) Ss were more likely to be residing in their natural homes than those cases in the remaining comparison population. Most potential cases from the original population were not included in either case group. Findings suggest that "convenience" samples differ significantly from their parent population and that caution is required when applying data generated from selected groups to the remaining population. (16 ref) (PsycINFO Database Record (c) 2016 APA, all rights reserved)},
issn={0022-264X(Print)}

@Article{Shikany2019,
author={Shikany, Amy R.
and Parrott, Ashley
and James, Jeanne
and Madueme, Peace
and Weaver, Kathryn Nicole
and Cassidy, Christine
and Khoury, Philip R.
and Miller, Erin M.},
title={Left ventricular outflow tract obstruction: Uptake of familial cardiac screening and parental knowledge from a single tertiary care center.},
journal={Journal of Genetic Counseling},
year={2019},
publisher={Wiley-Blackwell Publishing Ltd.},
address={Shikany, Amy R.: Heart Institute, Cincinnati Children's Hospital Medical Center, Cincinnati, OH, US, amy.shikany@cchmc.org},
volume={28},
pages={779-789},
keywords={*Cardiovascular Disorders; *Disease Screening; *Family; *Genetic Testing; *Health Care Services; Genetic Counseling; Genetics; Heritability; Screening Tests},
abstract={Left ventricular outflow tract obstruction (LVOTO) malformations exhibit higher heritability than other cardiac lesions and cardiac screening is encouraged for first-degree relatives. This study sought to determine the uptake of familial cardiac screening in families with an infant with an LVOTO and assess parental knowledge regarding genetics and heritability of LVOTO. A chart review of the

period 2010--2015 identified 69 families who received genetic counseling regarding a diagnosis of LVOTO in an infant. Surveys assessing familial cardiac screening and parental knowledge were completed by a parent in 24 families (completion rate of 35%). Forty percent (36/89) of all at-risk first-degree family members completed cardiac screening. The presence of additional congenital malformations in the affected infant was the only significant factor reducing the uptake of familial cardiac screening ($p = 0.003$). The reported uptake of screening for subsequent at-risk pregnancies was 11/12 (92%) compared to 25/77 (32%) of living at-risk relatives. Survey respondents answered seven knowledge questions with an average score of 5.2 and all correctly identified that LVOTO can run in families. Uptake of familial cardiac screening is occurring in less than half of at-risk individuals, despite parents demonstrating basic knowledge and receiving genetic counseling. Follow-up counseling in the outpatient setting to review familial screening recommendations should be considered to increase uptake and optimize outcomes. (PsycInfo Database Record (c) 2021 APA, all rights reserved)},
doi={10.1002/jgc4.1117},
url={https://doi.org/10.1002/jgc4.1117}

@Article{Southard2012,
author={Southard, Abigail E.
and Edelmann, Lisa J.
and Gelb, Bruce D.},
title={Role of copy number variants in structural birth defects.},
journal={Pediatrics},
year={2012},
publisher={American Academy of Pediatrics},
address={Gelb, Bruce D.: Child Health and Development Institute, Mount Sinai School of Medicine,
One Gustave Levy Place, Box 1040, New York, NY, US, 10029, bruce.gelb@mssm.edu},
volume={129},
pages={755-763},
keywords={*Birth; *DNA; *Genome; *Nucleotides; Polymorphism},
abstract={Background and Objective: Human genomes include copy number variants (CNVs), defined as regions with DNA gains or losses. Pathologic CNVs, which are larger and often occur de novo, are increasingly associated with disease. Given advances in genetic testing, namely microarray-based comparative genomic hybridization and single nucleotide polymorphism arrays, previously unidentified genotypic aberrations can now be correlated with phenotypic anomalies. The objective of this study was to conduct a nonsystematic literature review to document the role of CNVs as they relate to isolated structural anomalies of the craniofacial, respiratory, renal, and cardiac systems. Methods: All full-length articles in the PubMed database through May 2011 that discussed CNVs and isolated structural defects of the craniofacial, respiratory, renal, and cardiac systems were considered. Search terms queried include CNV, copy number variation, array comparative genomic hybridization, birth defects, craniofacial defects, respiratory defects, renal defects, and congenital heart disease. Reports published in languages other than English and articles regarding CNVs and neurocognitive deficits were not considered. Results: Evidence supports that putatively pathogenic CNVs occur at an increased frequency in patients with isolated structural birth defects and implicate specific regions of the genome. Through CNV detection, advances have been made in identifying genes and specific loci that underlie isolated birth defects. Conclusions: Although limited studies have been published, the promising evidence reviewed here warrants the continued investigation of CNVs in children with isolated structural birth defects. Patient care and genetic counseling stand to improve through a better understanding of CNVs and their effect on disease phenotype. (PsycINFO Database Record (c) 2016 APA, all rights reserved)},
doi={10.1542/peds.2011-2337},

url={<https://doi.org/10.1542/peds.2011-2337>}

@Article{Streissguth1977,
author={Streissguth, Ann Pytkowicz},
title={Maternal drinking and the outcome of pregnancy: Implications for child mental health.},
journal={American Journal of Orthopsychiatry},
year={1977},
publisher={American Orthopsychiatric Association, Inc.},
address={Streissguth, Ann Pytkowicz: Division of Child Psychiatry, University of Washington, Mail Stop GI-80, Seattle, WA, US, 98195},
volume={47},
pages={422-431},
keywords={*At Risk Populations; *Fetal Alcohol Syndrome; *Mental Health; *Pregnancy; *Risk Factors; Clinics; Human Females; Offspring},
abstract={Research since identification in 1973 of the fetal alcohol syndrome indicates that offspring of alcoholic women who drink heavily during pregnancy are at high risk for physical and mental deficiencies, and that even "social drinking" during pregnancy may have detrimental effects on birthweight and behavior of infants. While further research and remediation efforts are needed, primary prevention and active intervention in counseling and obstetrical clinics seem clearly and urgently called for. (PsycINFO Database Record (c) 2019 APA, all rights reserved)},
doi={10.1111/j.1939-0025.1977.tb01248.x},
url={<https://doi.org/10.1111/j.1939-0025.1977.tb01248.x>}

@Inbook{Liamlahi2019,
author={Liamlahi, Rabia
and Latal, Beatrice},
title={Neurodevelopmental outcome of children with congenital heart disease.},
bookTitle={Neonatal neurology.},
series={Handbook of clinical neurology.},
year={2019},
publisher={Elsevier},
address={Latal, Beatrice: Child Development Center, University Children's Hospital Zurich, Steinwiesstrasse 75, Zurich, Switzerland, 8032, bea.latal@kispi.uzh.ch},
pages={329-345},
keywords={*Brain Development; *Congenital Disorders; *Heart Disorders; *Neurodevelopmental Disorders; *Pediatrics; Etiology},
abstract={Congenital heart disease (CHD) constitutes the most common congenital malformation, with moderate or severe CHD occurring in around 6 in 1000 live births. Due to advances in medical care, survival rates have increased significantly. Thus, the majority of children with CHD survive until adolescence and adulthood. Children with CHD requiring cardiopulmonary bypass surgery are at risk for neurodevelopmental impairments in various domains, including mild impairments in cognitive and neuromotor functions, difficulties with social interaction, inattention, emotional symptoms, and impaired executive function. The prevalence for these impairments ranges from 20{\%} to 60{\%} depending on age and domain ("high prevalence--low severity"). Domains are often affected simultaneously, leading to school problems with the need for learning support and special interventions. The etiology of neurodevelopmental impairments is complex, consisting of a combination of delayed intrauterine brain development and newly occurring perioperative brain injuries. Mechanisms include altered intrauterine hemodynamic flow as well as neonatal hypoxia and reduced cerebral blood flow. The surgical

procedure and postoperative phase add to this cascade of factors interfering with normal brain development. Early identification of children at high risk through structured follow-up programs is mandated to provide individually tailored early interventions and counseling to improve developmental health. (PsycINFO Database Record (c) 2019 APA, all rights reserved)),
isbn={978-0-444-64029-1 (Hardcover)},
doi={10.1016/B978-0-444-64029-1.00016-3},
url={https://doi.org/10.1016/B978-0-444-64029-1.00016-3}

@Article{Newall2008,
author={Newall, Fiona
and Johnston, Linda
and Monagle, Paul},
title={Optimising anticoagulant education in the paediatric setting using a validated model of education.},
journal={Patient Education and Counseling},
year={2008},
publisher={Elsevier Science},
address={Newall, Fiona: Clinical Haematology Department, Royal Children's Hospital, 9th Floor, Main Building, Flemington Road, Parkville, VIC, Australia, 3052, fiona.newall@rch.org.au},
volume={73},
pages={384-388},
keywords={*Anticoagulant Drugs; *Client Education; *Health Knowledge; *Intervention; Drug Therapy; Pediatrics},
abstract={Objective: Providing education to patients requiring anticoagulant therapy may be associated with improved outcomes. This study investigated the knowledge outcomes of a validated educational intervention. Methods: Parents of children with congenital heart disease requiring warfarin therapy took part in an educational intervention. Warfarin knowledge was assessed prior to commencing the program, immediately following its completion and 6 months following completion. Results: Parents demonstrated a statistically significant improvement in their warfarin knowledge immediately following completion of the program ($p < 0.0001$), with this improvement being sustained over time. Conclusion: Current approaches to educating parents of children requiring warfarin therapy are likely suboptimal. Using a validated model of education may be associated with improved knowledge outcomes for patients. Practice Implications: Consideration to the processes used in delivering patient education may result in improved patient knowledge outcomes. (PsycInfo Database Record (c) 2020 APA, all rights reserved)},
doi={10.1016/j.pec.2008.07.027},
url={https://doi.org/10.1016/j.pec.2008.07.027}}

@Article{Hancock2022,
author={Hancock, Bailey
and Miller, Erin M.
and Parrott, Ashley
and Weaver, Kathryn Nicole
and Tretter, Justin T.
and Pilipenko, Valentina
and Shikany, Amy R.},

title={Retrospective comparison of parent-reported genetics knowledge, empowerment, and familial uptake of cardiac screening between parents who received genetic counseling by a certified genetic counselor and those who did not: A single us academic medical cent},
journal={Journal of Genetic Counseling},
year={2022},
publisher={Wiley-Blackwell Publishing Ltd.},
address={United Kingdom},
pages={No Pagination Specified-No Pagination Specified},
abstract={Bicuspid aortic valve (BAV) is the most common congenital heart defect, which can cause severe cardiac complications. BAVs cluster in families and demonstrate high heritability. Cardiac screening for first-degree relatives of individuals with a BAV is recommended. This retrospective two-group study evaluated the impact of cardiovascular genetic counseling provided by a board-certified genetic counselor on parent-reported outcomes by comparing parental responses of those who received genetic counseling by a genetic counselor (GC group) for family history of BAV to those who did not (non-GC group). A retrospective chart review from May 2016 to June 2019 identified 133 pediatric patients with an isolated BAV. Parents of eligible probands were invited to complete an online survey assessing genetics knowledge, empowerment (Genomics Outcome Scale), and familial uptake of cardiac screening. Surveys were completed by 38/97 (39%) parents in the non-GC group and 20/36 (56%) parents in the GC group. The median genetics knowledge score was not significantly different between the two groups (GC group: 8, range 3--11 out of a maximum possible of 12; non-GC group: 7, range 2--11; $p = .08$). The mean empowerment score was not significantly different between the two groups (GC group: mean 24.6, SD 2.2; non-GC group: mean 23.2, SD 3.5; $p = .06$). The uptake of cardiac screening was significantly higher in the GC group with 39/59 (66%) total first-degree relatives reported as having been screened compared with 36/91 (40%) in the non-GC group ($p = .002$). Parent-reported outcomes in our study suggest that receiving genetic counseling by a board-certified genetic counselor significantly increased familial uptake of cardiac screening for first-degree relatives of pediatric patients with a BAV. Studies with larger sample sizes are needed to confirm the findings of this study; however, a referral to a genetic counselor should be considered for patients with a BAV. (PsycInfo Database Record (c) 2022 APA, all rights reserved)},
doi={10.1002/jgc4.1570},
url={https://doi.org/10.1002/jgc4.1570}

@PhdThesis{Simko2000,
author={Simko, Lynn Marie Coletta},
title={Quality of life of adults with congenital heart disease.},
year={2000},
publisher={ProQuest Information {&} Learning},
address={US},
volume={61},
pages={1329-1329},
keywords={*Congenital Disorders; *Counseling; *Heart Disorders; Quality of Life},
abstract={Adults with Congenital Heart Disease (CHD) are a new and growing population of patients who pose a challenge to both medicine and nursing. Medical and surgical advances have increased the number of adults with CHD. These advances may create Quality of Life (QOL) issues which were not previously considered. The purpose of this study was to collect data describing the QOL of adults with CHD in order for health professionals to develop appropriate information, counseling, and anticipatory guidance. This study was a case-control study design utilizing a QOL assessment tool, the Sickness impact Profile (SIP), along with a demographic questionnaire. A convenience sample of 124 adults with CHD and 124 matched healthy controls participated in the study. Overall, SIP scores and

each of the component subscores range from 0 (no disability) to 100. There was a statistically significant difference between the adults with CHD and their healthy controls in the total SIP score, the physical and psychosocial dimension score and all category scores ($p < .05$). The areas of life of the adults with CHD reported as being most affected were the category scores of work (SIP 11.1), and sleep and rest (SIP of 9.03). The data did not reveal a statistically significant difference between those with cyanotic anomalies versus acyanotic anomalies, however when those individuals with Tricuspid Atresia (TAT) and Single Ventricle (SIV) were combined, there was a statistically significant difference between TAT and SIV and those with acyanotic anomalies ($p < .05$). The primary diagnosis of those adults reporting the poorest QOL was TAT. Total SIP scores for adults with TAT were statistically significantly higher ($p < .05$) than those adults with the primary diagnoses of coarctation of the aorta (COA) and aortic stenosis, general (ASG), and borderline statistically significant difference ($p < .10$) between those with TAT and pulmonic stenosis, general (PSG). The significance of this study does indicate that adults with CHD did not see themselves as having any physical limitations, their perception of themselves and their individual expectations are normal. (PsycINFO Database Record (c) 2016 APA, all rights reserved)},
issn={0419-4217(Print)}

@Article{Helm2018,
author={Helm, Benjamin M.
and Freeze, Samantha L.
and Spoonamore, Katherine G.
and Ware, Stephanie M.
and Ayers, Mark D.
and Kean, Adam C.},
title={The genetic counselor in the pediatric arrhythmia clinic: Review and assessment of services.},
journal={Journal of Genetic Counseling},
year={2018},
publisher={Springer},
address={Helm, Benjamin M.: Department of Medical & Molecular Genetics, Indiana University School of Medicine, Riley Hospital for Children, 550 N. University Blvd., AOC 5th Floor, Ste. 5051, Indianapolis, IN, US, 46202, bmhelm@iu.edu},
volume={27},
pages={558-564},
keywords={*Arrhythmias (Heart); *Electrophysiology; *Genetic Counseling; *Heart Disorders; *Sudden Death; Pediatrics},
abstract={There are minimal data on the impact of genetic counselors in subspecialty clinics, including the pediatric arrhythmia clinic. This study aimed to describe the clinical encounters of a genetic counselor integrated into a pediatric arrhythmia clinic. In the 20 months between July 2015 and February 2017, a total of 1914 scheduled patients were screened for indications relevant for assessment by a genetic counselor. Of these, the genetic counselor completed 276 patient encounters, seeing 14.4% of all patients in clinic. The most expected and common indications for genetic counselor involvement were related to suspicion for primary heritable arrhythmia conditions, though patients seen in this clinic display a wide range of cardiac problems and many additional indications for genetic evaluation were identified. Roughly 75% (211/276) of encounters were for personal history of confirmed/suspected heritable disease, including cardiac channelopathies, cardiomyopathies, ventricular arrhythmias, and congenital heart defects, and 25% (65/276) were for family history of disease, including long QT syndrome and sudden unexplained death. Overall, this study shows that about 1 in 7 patients seen in a pediatric arrhythmia clinic have indications that likely benefit from genetic counselor involvement and care. Similar service delivery models embedding genetic counselors in pediatric arrhythmia clinics should be encouraged, and this model could be

emulated to increase patient access to genetic counseling services. (PsycINFO Database Record (c) 2019 APA, all rights reserved)),
doi={10.1007/s10897-017-0169-5},
url={https://doi.org/10.1007/s10897-017-0169-5}

@Article{Simko2006,
author={Simko, Lynn Coletta
and McGinnis, Kathleen A.
and Schembri, Joanne},
title={Educational needs of adults with congenital heart disease.},
journal={Journal of Cardiovascular Nursing},
year={2006},
publisher={Lippincott Williams {&} Wilkins},
address={Simko, Lynn Coletta: School of Nursing, Duquesne University, Pittsburgh, PA, US, 15282,
Simko@duq.edu},
volume={21},
pages={85-94},
keywords={*Congenital Disorders; *Health Education; *Heart Disorders; Needs Assessment; Nursing},
abstract={Adults with congenital heart disease represent a new and growing population of patients who pose a challenge to both medicine and nursing. The purpose of this study was to identify the educational needs of adults with congenital heart disease. The study used a prospective, cross-sectional, case-control study design and enrolled 124 adults with congenital heart disease and 124 matched healthy controls. Adults with congenital heart disease were more likely to report living at home with parents and less likely to be employed compared with healthy controls. When those adults with acyanotic and cyanotic congenital heart disease were compared, acyanotic adults were twice as likely to be employed. Additionally, several areas of educational needs were identified in this study for adults with congenital heart disease. (PsycINFO Database Record (c) 2018 APA, all rights reserved)),
doi={10.1097/00005082-200603000-00003},
url={https://doi.org/10.1097/00005082-200603000-00003}}

@Article{Goldberg1974,
author={Goldberg, Richard T.},
title={Adjustment of children with invisible and visible handicaps: Congenital heart disease and facial burns.},
journal={Journal of Counseling Psychology},
year={1974},
publisher={American Psychological Association},
address={US},
volume={21},
pages={428-432},
keywords={*Burns; *Heart Disorders; *Psychosocial Development; Social Adjustment},
abstract={26 11-15 yr old children with congenital heart disease and 22 with facial burns were compared on 10 measures of adjustment to test the effects of invisible and visible disability upon social and psychological development. Interview data were coded and submitted to analysis of covariance, using sex, grade, and age as covariates. The invisible disability group (the heart group) was higher in adjustment in all 10 measures. Significant differences ($p < .01$) were obtained on vocational aspirations, origin of interest in vocational aspiration, career plans after high school, self-image, and work values. It is concluded that an invisible disability with severe physical limitations

has fewer deleterious effects upon social adjustment than does a visible disability without physical limitations such as facial disfigurement. (20 ref) (PsycINFO Database Record (c) 2016 APA, all rights reserved)),

doi={10.1037/h0037086},

url={https://doi.org/10.1037/h0037086}

@Article{Morton2011,

author={Morton, Liza},

title={Can Interpersonal Psychotherapy (IPT) meet the psychosocial cost of life gifted by medical intervention?},

journal={Counselling Psychology Review},

year={2011},

publisher={British Psychological Society},

address={Morton, Liza: Department of Clinical Psychology, Lynebank Hospital, Halbeath Road, Dunfermline, United Kingdom, KY11 4UW, lizamorton@nhs.net},

volume={26},

pages={75-86},

keywords={*Congenital Disorders; *Emotional Adjustment; *Heart Disorders; *Interpersonal Psychotherapy; *Well Being; Survivors},

abstract={Context and focus: Advances in medicine and technology have improved the survival of children born with life threatening medical conditions. However, this gift of life entails potential psychological, emotional and social costs including the impact of enduring paediatric and adult medical trauma, growing up feeling different, tolerating discrimination and disempowerment and adjusting to a further loss of health. Here, these emotional issues are explored using congenital heart disease as a specific example. Currently, psychological difficulties are under diagnosed for this population which could impact not only on psychological well-being and quality of life but also recovery from illness. As such, the potential benefits of Interpersonal Psychotherapy (IPT) as a treatment option are explored. Conclusions: IPT seems a suitable treatment option for this population. Given that many of the issues faced by congenital heart disease survivors are likely to be shared with individuals born with other congenital conditions it seems likely that IPT generally offers a suitable treatment option for adults with a lifelong illness. A focus on the interpersonal difficulties faced by such individuals could benefit clients by validating the psychosocial cost that these individuals pay to live and endeavouring to develop an interpersonal network that better meets their complex psychological and emotional needs. Some potential limitations are discussed. (PsycINFO Database Record (c) 2019 APA, all rights reserved)}

@Article{Wade2020,

author={Wade, Shari L.

and Gies, Lisa M.

and Fisher, Allison P.

and Moscato, Emily L.

and Adlam, Anna R.

and Bardoni, Alessandra

and Corti, Claudia

and Limond, Jennifer

and Modi, Avani C.

and Williams, Tricia},

title={Telepsychotherapy with children and families: Lessons gleaned from two decades of translational research.},
journal={Journal of Psychotherapy Integration},
year={2020},
publisher={Educational Publishing Foundation},
address={Wade, Shari L.: Division of Pediatric Rehabilitation Medicine, Cincinnati Children's Hospital Medical Center, 3333 Burnet Avenue, MLC 4009, Cincinnati, OH, US, 45229,
shari.wade@cchmc.org},
volume={30},
pages={332-347},
keywords={*Epilepsy; *Parent Training; *Psychotherapy; *Telemedicine; *Traumatic Brain Injury; Cerebrovascular Accidents; Clinical Practice; Family; Heart Disorders; Intervention; Neoplasms; Problem Solving; Treatment Effectiveness Evaluation; Viral Disorders},
abstract={The novel coronavirus, COVID-19, has led to sweeping changes in psychological practice and the concomitant rapid uptake of telepsychotherapy. Although telepsychotherapy is new to many clinical psychologists, there is considerable research on telepsychotherapy treatments. Nearly 2 decades of clinical research on telepsychotherapy treatments with children with neurological conditions has the potential to inform emerging clinical practice in the age of COVID-19. Toward that end, we synthesized findings from 14 clinical trials of telepsychotherapy problem-solving and parent-training interventions involving more than 800 children and families with diverse diagnoses, including traumatic brain injury, epilepsy, brain tumors, congenital heart disease, and perinatal stroke. We summarize efficacy across studies and clinical populations and report feasibility and acceptability data from the perspectives of parents, children, and psychotherapists. We describe adaptation for international contexts and strategies for troubleshooting technological challenges and working with families of varying socioeconomic strata. The extensive research literature reviewed and synthesized provides considerable support for the utility of telepsychotherapy with children with neurological conditions and their families and underscores its high level of acceptability with both diverse clinical populations and providers. During this period of heightened vulnerability and stress and reduced access to usual supports and services, telepsychotherapy approaches such as online family problem-solving treatment and online parenting skills training may allow psychologists to deliver traditional evidence-based treatments virtually while preserving fidelity and efficacy. (Psychnfo Database Record (c) 2021 APA, all rights reserved)},
doi={10.1037/int0000215},
url={https://doi.org/10.1037/int0000215}

@Article{Morales2017a,
author={Morales, Ana
and Allain, Dawn C.
and Arscott, Patricia
and James, Emily
and MacCarrick, Gretchen
and Murray, Brittney
and Tichnell, Crystal
and Shikany, Amy R.
and Spencer, Sara
and Fitzgerald-Butt, Sara M.
and Kushner, Jessica D.
and Munn, Christi
and Smith, Emily
and Spoonamore, Katherine G.

and Tandri, Harikrishna S.
and Kay, W. Aaron},
title={"At the heart of the pregnancy: What prenatal and cardiovascular genetic counselors need to know about maternal heart disease": Erratum.},
journal={Journal of Genetic Counseling},
year={2017},
publisher={Springer},
address={Morales, Ana: Human Genetics Division, Ohio State University, 306 BRT, 460 W. 12th Ave, Columbus, OH, US, 43210, ana.morales@osumc.edu},
volume={26},
pages={689-689},
keywords={*Counselors; *Genetics; *Heart Disorders; *Knowledge Level; *Pregnancy; Mothers; Prenatal Development},
abstract={Reports an error in "At the heart of the pregnancy: What prenatal and cardiovascular genetic counselors need to know about maternal heart disease" by Ana Morales, Dawn C. Allain, Patricia Arscott, Emily James, Gretchen MacCarrick, Brittney Murray, Crystal Tichnell, Amy R. Shikany, Sara Spencer, Sara M. Fitzgerald-Butt, Jessica D. Kushner, Christi Munn, Emily Smith, Katherine G. Spoonamore, Harikrishna S. Tandri and W. Aaron Kay (Journal of Genetic Counseling, Advanced Online Publication, Mar 10, 2017, np). The original version of this article contained some mistakes in Tables 1 and 3. The corrections are provided in the erratum. (The following abstract of the original article appeared in record 2017-11495-001). In the last decade, an increasing number of cardiac conditions have been shown to have a genetic basis. Cardiovascular genetic counseling has emerged as a subspecialty aiming to identify unaffected at-risk individuals. An important sector of this at-risk population also includes expectant mothers, in whom unique clinical challenges may arise. Genetic counselors, especially those in cardiovascular and prenatal settings, have an opportunity to identify and assist women who may benefit from cardiovascular care during pregnancy. This paper provides basic management and genetic evaluation principles for affected women, as well as guidance on identifying those who are at risk. We provide considerations for cardiac surveillance in pregnancy and the post-partum period. Finally, key psychosocial issues that appraise how to best provide support to at risk women as they make informed decisions are discussed. We propose that a team approach including cardiology, maternal fetal medicine, and genetic counseling best serves this patient population. Ongoing questions addressing an evidence based approach to cardiovascular genetic conditions in pregnancy still remain. Thus, well-designed research protocols are essential to mark progress in this area. (PsycINFO Database Record (c) 2019 APA, all rights reserved)},
doi={10.1007/s10897-017-0101-z},
url={https://doi.org/10.1007/s10897-017-0101-z}

@Article{Mangset2014,
author={Mangset, Margrete
and Hofmann, Bj{\o}rn},
title={LQTS parents' reflections about genetic risk knowledge and their need to know or not to know their children's carrier status.},
journal={Journal of Genetic Counseling},
year={2014},
publisher={Springer},
address={Mangset, Margrete: University of Oslo, Faculty of Medicine, Institute of Health and Society, Centre for Medical Ethics, Postboks 1130, Blindern, Oslo, Norway, 0318,
margrete.mangset@medisin.uio.no},
volume={23},
pages={1022-1033},

keywords={*Genetic Counseling; *Genetic Disorders; *Parent Child Relations; *Parents; *Risk Factors; Autonomy; DNA; Genetics; Independence (Personality); Responsibility},
abstract={Long QT syndrome (LQTS) is a contributor to unexplained deaths in infants (SIDS), children, teenagers and young adults. A gene test result may allow for individual tailored treatment, but also pose a burden of knowing one's carrier status, with no treatment recommendation. Genetic risk knowledge in the case of LQTS can promote adjustment and coping, but also fear anxiety, ambivalence and moral dilemmas. This makes it challenging to respect both the right to know and the right not to know. The purpose of this study was to explore LQTS parents' perception of genetic knowledge, and their need to know or not to know about their children's carrier status. Qualitative, semi structured interviews were conducted with thirteen parents of LQTS-children. Results show that parents found it important to know the result of a gene test for LQTS including their children's carrier status. The risk was framed and incorporated into their everyday life and their life perspectives. Pertinent moral dilemmas concerned information disclosure to children and relatives. Parents thought that early and gradual disclosure to children would promote coping. Parents' moral dilemmas were rarely addressed during encounters with healthcare providers. The participants had several suggestions for improvement in that regard. (PsycINFO Database Record (c) 2019 APA, all rights reserved)},
doi={10.1007/s10897-014-9727-2},
url={https://doi.org/10.1007/s10897-014-9727-2}

@Article{Morales2017b,
author={Morales, Ana
and Allain, Dawn C.
and Arscott, Patricia
and James, Emily
and MacCarrick, Gretchen
and Murray, Brittney
and Tichnell, Crystal
and Shikany, Amy R.
and Spencer, Sara
and Fitzgerald-Butt, Sara M.
and Kushner, Jessica D.
and Munn, Christi
and Smith, Emily
and Spoonamore, Katherine G.
and Tandri, Harikrishna S.
and Kay, W. Aaron},
title={At the heart of the pregnancy: What prenatal and cardiovascular genetic counselors need to know about maternal heart disease.},
journal={Journal of Genetic Counseling},
year={2017},
publisher={Springer},
address={Morales, Ana: Human Genetics Division, Ohio State University, 306 BRT, 460 W. 12th Ave, Columbus, OH, US, 43210, ana.morales@osumc.edu},
volume={26},
pages={669-688},
keywords={*Counselors; *Genetics; *Heart Disorders; *Knowledge Level; *Pregnancy; Genetic Counseling; Mothers; Prenatal Development},
abstract={Correction Notice: An Erratum for this article was reported in Vol 26(4) of Journal of Genetic Counseling (see record 2017-30007-001). The original version of this article contained some mistakes

in Tables 1 and 3. The corrections are provided in the erratum.] In the last decade, an increasing number of cardiac conditions have been shown to have a genetic basis. Cardiovascular genetic counseling has emerged as a subspecialty aiming to identify unaffected at-risk individuals. An important sector of this at-risk population also includes expectant mothers, in whom unique clinical challenges may arise. Genetic counselors, especially those in cardiovascular and prenatal settings, have an opportunity to identify and assist women who may benefit from cardiovascular care during pregnancy. This paper provides basic management and genetic evaluation principles for affected women, as well as guidance on identifying those who are at risk. We provide considerations for cardiac surveillance in pregnancy and the post-partum period. Finally, key psychosocial issues that appraise how to best provide support to at risk women as they make informed decisions are discussed. We propose that a team approach including cardiology, maternal fetal medicine, and genetic counseling best serves this patient population. Ongoing questions addressing an evidence based approach to cardiovascular genetic conditions in pregnancy still remain. Thus, well-designed research protocols are essential to mark progress in this area. (PsycINFO Database Record (c) 2019 APA, all rights reserved)),
doi={10.1007/s10897-017-0081-z},
url={https://doi.org/10.1007/s10897-017-0081-z}

@Article{Werner2019,
author={Werner, Oscar
and Abassi, Hamouda
and Lavastre, Kathleen
and Guillaumont, Sophie
and Picot, Marie-Christine
and Serrand, Chris
and Dulac, Yves
and Souletie, Nathalie
and Acar, Philippe
and Bredy, Charlene
and Amedro, Pascal},
title={Factors influencing the participation of adolescents and young adults with a congenital heart disease in a transition education program: A prospective multicentre controlled study.},
journal={Patient Education and Counseling},
year={2019},
publisher={Elsevier Science},
address={Amedro, Pascal: Paediatric and Congenital Cardiology Department, Montpellier University Hospital, 371 Avenue du Doyen Giraud, Montpellier, France, 34295, p-amedro@chu-montpellier.fr},
volume={102},
pages={2223-2230},
keywords={*Client Education; *Client Participation; *Heart Disorders; *Life Changes; *Quality of Life; Autonomy; Cardiology},
abstract={Objective: Transition education programs dedicated to adolescents and young adults with congenital heart disease (CHD) aim to facilitate transfer to adult cardiology and bring more autonomy to teenagers. This prospective controlled multicentre study analysed the factors influencing the participation in a transition education program. Methods: CHD patients aged 13--25 y were offered to participate in the transition program. A multiple linear regression identified the explanatory factors for participation in the program. Results: A total of 123 patients (mean age 19.6 {textpm} 3.4 y) were included in the study, with 57 participants and 66 non-participants. Both groups showed similar socio-demographic and quality of life characteristics, low level of physical activity with muscular deconditioning and high exposure to risk behaviours (71{\%} patients with ≥ 1 risk factor). Patients with

complex CHD (OR = 4.1, P = 0.03), poor disease knowledge (OR = 0.3, P = 0.02), risk behaviours (body piercing, OR = 5.53, P = 0.01; alcohol, OR = 3.12, P = 0.06), and aged < 20 y (OR = 0.29, P = 0.03), were more likely to join the program. Conclusion: Many risk factors influencing the participation of adolescents and young adults with CHD in transition education programs are controllable. Practice implication: Further randomized studies are necessary to evaluate the impact of transition education program on quality of life, successful transfer to adult centre and, ultimately, prognosis. (PsycInfo Database Record (c) 2020 APA, all rights reserved)),
doi={10.1016/j.pec.2019.06.023},
url={https://doi.org/10.1016/j.pec.2019.06.023}

@Article{Kannai2012,
author={Kannai, Ruth},
title={`Jum'ah" syndrome.},
journal={Patient Education and Counseling},
year={2012},
publisher={Elsevier Science},
address={Kannai, Ruth: Department of Family Medicine, Hebrew University, Hadassah School of Medicine, P.O. Box 12272, 6B Apt. 2 Reuven St., Bet Shemesh, Jerusalem, Israel, 99544,
ruthyka@inter.net.il},
volume={87},
pages={133-134},
keywords={*Anxiety; *Hospitalization; *Hospitals; Pediatrics},
abstract={This is my memoir as a sick child, hospitalized in the Pediatric ward of a large hospital, many years ago. The story tells about my friendship with another young patient, Jum'ah, a Bedouin child who suffered from Congenital Cyanotic Heart Disease, to whom the pediatric ward was Home. My Childish understanding of Jum'ah's loneliness, anxiety and struggle to be loved and belonged are described in this narrative. I describe how this experience still has an influence on my adult professional and personal concepts. (PsycInfo Database Record (c) 2020 APA, all rights reserved)),
doi={10.1016/j.pec.2011.08.018},
url={https://doi.org/10.1016/j.pec.2011.08.018}

@Article{Walter1992,
author={Walter, P. J.
and Mohan, R.
and Dahan-Mizrahl, S.},
title={Quality of life after open heart surgery 16--28 May 1991.},
journal={Quality of Life Research: An International Journal of Quality of Life Aspects of Treatment, Care {&} Rehabilitation},
year={1992},
publisher={Springer},
address={Germany},
volume={1},
pages={77-83},
keywords={*Heart Surgery; Quality of Life},
abstract={Reviews papers presented at the international symposium on "Quality of Life after Open Heart Surgery" comprising the experiences of more than 20,000 patients. Early identification and operation of premorbid personalities, psychological counseling before and after the operation, and formulating socioeconomic policies that encourage returning to work yielded improvement in all patient

groups. Specific to valve replacement was patient participation in deciding the kind of prosthesis and improvement of anticoagulation strategies. Coronary bypass patients benefited through a comprehensive rehabilitation program. For congenital heart disease, emphasis was laid on the need to educate parents about the disease; the importance of open discussion between parents, the patient, and the pediatric cardiologist; and the function of a parents' self-help group. For heart transplantation, the endeavor must be to increase donor availability and to develop better immunosuppression schedules. (PsycINFO Database Record (c) 2016 APA, all rights reserved)),
doi={10.1007/BF00435439},
url={https://doi.org/10.1007/BF00435439}

@Article{Bertaud2020,
author={Bertaud, Sophie
and Lloyd, David F. A.
and Sharland, Gurleen
and Razavi, Reza
and Bluebond-Langner, Myra},
title={The impact of prenatal counselling on mothers of surviving children with hypoplastic left heart syndrome: A qualitative interview study.},
journal={Health Expectations: An International Journal of Public Participation in Health Care {&} Health Policy},
year={2020},
publisher={Wiley-Blackwell Publishing Ltd.},
address={Bertaud, Sophie: Louis Dundas Centre for Children's Palliative Care, Great Ormond Street Hospital, Barclay House, Great Ormond Street, London, United Kingdom, WC1N 3JH, sophie.beraud@gosh.nhs.uk},
volume={23},
pages={1224-1230},
keywords={*Antepartum Period; *Counseling; *Fetus; *Heart Disorders; *Mothers; Caregivers; Diagnosis; Parents},
abstract={Objective: To explore the role of antenatal counselling in how parents make treatment decisions following an antenatal diagnosis of Hypoplastic Left Heart Syndrome (HLHS). Background: Antenatal counselling is a critical part of patient management following a diagnosis of fetal congenital heart disease; however, there is a very limited evidence base examining how parents actually experience antenatal counselling and make decisions in this context. Methods: Semi-structured interviews were conducted with women who had received an antenatal diagnosis of HLHS. Interviews were digitally recorded, anonymised and transcribed verbatim. A thematic content analysis was performed using a constant comparative approach. Results: Eight mothers of surviving children with HLHS were interviewed. Eight key themes emerged including new perspectives on how women receive antenatal counselling and how it affects their decision making. Three themes in particular are new to the literature: (a) Mothers of children with HLHS reported feelings of intense guilt that arose in the antenatal period around potentially causing the condition in their child. (b) For this group of women, recollections of perceived pessimism during antenatal counselling had a lasting impact. (c) Despite support from partners or extended family, women nevertheless experienced a strong sense that antenatal decision making was largely a 'maternal' responsibility. Conclusions: When recounting their experiences of antenatal counselling, mothers of surviving children with HLHS offer new perspectives that can guide fetal cardiologists in how best to support their individual patients. Further research is needed to comprehensively understand the experience of prospective parents counselled for severe forms of fetal congenital heart disease. (PsycInfo Database Record (c) 2021 APA, all rights reserved)),
doi={10.1111/hex.13103},

url={https://doi.org/10.1111/hex.13103}

@Article{Li2018,
author={Li, Yaxi
and Solomon, Phyllis
and Zhang, Anao
and Franklin, Cynthia
and Ji, Qingying
and Chen, Yuting},
title={Efficacy of solution-focused brief therapy for distress among parents of children with congenital heart disease in China.},
journal={Health {&} Social Work},
year={2018},
publisher={Oxford University Press},
address={Ji, Qingying: Shanghai Children's Medical Center, Shanghai, China, 200127,
jiquingying@scmc.com.cn},
volume={43},
pages={30-39},
keywords={*Brief Psychotherapy; *Congenital Disorders; *Distress; *Heart Disorders; *Solution Focused Therapy; Parents},
abstract={Chinese parents of children with congenital heart disease (CHD) experience significant psychological distress due to the child's illness and hospitalization. Unfortunately, there are few psychosocial interventions for parental distress in China. This pilot study aimed to examine the efficacy of solution-focused brief therapy (SFBT) in a Chinese hospital for parental distress using a randomized controlled trial design. The participants included 40 Chinese parents of a currently hospitalized child diagnosed with CHD who were assessed to have psychological distress. Parents were randomized into either the intervention (n = 25) or the hospital medical social work treatment as usual (TAU) (n = 28) group. The Chinese Brief Symptom Inventory-18 and Chinese version of Herth Hope Index were administered before and after the interventions. Results of the intent-to-treat analysis indicated a significant decrease in parental distress and increase in parents' levels of hope in the intervention group compared with the TAU group. This study supported SFBT administered in a hospital setting as a promising intervention for reducing distress among Chinese parents with children diagnosed with CHD. (PsycInfo Database Record (c) 2022 APA, all rights reserved)},
doi={10.1093/hsw/hlx045},
url={https://doi.org/10.1093/hsw/hlx045}

@Article{MacMillin2003,
author={MacMillin, Martha D.},
title={Genetic library.},
journal={Journal of Genetic Counseling},
year={2003},
publisher={Springer},
address={MacMillin, Martha D.: 11 Briar Hill Road, Sharon, MA, US, 02067,
mmacmillin@netscape.net},
volume={12},
pages={373-379},
keywords={*Genetic Counseling; *Genetic Testing; *Genetics; Physical Disorders},

abstract={Provides brief reviews/summaries of 8 articles presenting research in the field of genetic counseling. The first 3 articles are from a special issue of the Journal of Health Psychology and focus on the use of interpretative phenomenological analysis in the examination of psychological/social issues in the "new genetics" (clinical advances of the human genome project). Article 1 provides an overview of IPA, while articles 2 and 3 examine patients' perceptions of quality-of-life issues when living with an early/late-onset disease and an effective genetic counseling session. The following 6 articles involve preferences for knowing doctors' opinions among women considering clinical testing for BRCA1/2 mutations; genetic counseling for congenital heart disease; genetic counseling for sex chromosome abnormalities; outcomes of pregnancies diagnosed with Klinefelter syndrome; couples presenting for preimplantation genetic diagnosis; and genetics and metabolic causes of stillbirth. (PsycINFO Database Record (c) 2019 APA, all rights reserved)},
doi={10.1023/A:1023957123198},
url={https://doi.org/10.1023/A:1023957123198}

@Article{Wynn2018,
author={Wynn, Julia
and Martinez, Josue
and Bulafka, Jessica
and Duong, Jimmy
and Zhang, Yuan
and Chiuzan, Codruta
and Preti, Jain
and Cremona, Maria L.
and Jobanputra, Vaidehi
and Fyer, Abby J.
and Klitzman, Robert L.
and Appelbaum, Paul S.
and Chung, Wendy K.},
title={Impact of receiving secondary results from genomic research: A 12-month longitudinal study.},
journal={Journal of Genetic Counseling},
year={2018},
publisher={Springer},
address={Chung, Wendy K.: Department of Medicine, Columbia University Medical Center, 1150 St. Nicholas Ave., Russ Berrie Pavilion, 6th Fl, Rm, 620, New York, NY, US, wkc15@columbia.edu},
volume={27},
pages={709-722},
keywords={*Genetic Counseling; *Genome; *Genomic Sequencing; *Psychosocial Factors; Test Construction},
abstract={The impact of returning secondary results from exome sequencing (ES) on patients/participants is important to understand as ES is increasingly utilized in clinical care and research. Participants were recruited from studies using ES and were separated into two arms: 107 who had ES and were offered the choice to learn secondary results (ES group) and 85 who had not yet had ES (No ES group). Questionnaires were administered at baseline and 1 and 12 months, following results disclosure (ES group) or enrollment (No ES group). While the majority (65{\%}) elected to learn all results following pre-test counseling, it was reduced from the 76{\%} who indicated a desire for all results at baseline. Thirty-seven percent received results associated with an increased personal disease risk. There were no differences in changes in any of the psychological and social measures from baseline to post-results disclosure between the ES and No ES groups. Receiving a wide range of secondary findings appeared to have little measurable impact on most participants. The experience of learning secondary results may be related to participants' previous experiences with

genetics, as well as the genetic counseling provided. Future research with a more diverse, genetically naive group, as well as scalable methods of delivery, is needed. (PsycINFO Database Record (c) 2019 APA, all rights reserved)),
doi={10.1007/s10897-017-0172-x},
url={https://doi.org/10.1007/s10897-017-0172-x}

@Book{Money1990,
author={Money, John},
title={Four tutorials in pediatric sexology.},
series={Childhood and adolescent sexology.},
year={1990},
publisher={Elsevier Science},
address={New York, NY, US},
pages={137-168},
keywords={*Gender Identity; *Genital Disorders; *Psychosexual Behavior; Congenital Disorders; Heterosexuality; Homosexuality; Psychosexual Development; Sexology; Sexual Abuse},
abstract={[[tutorial 1] sex and eros: birth to age three [prenatal sexuality; genital hygiene; grooming sensuousness; nudity; parental intercourse; anatomical naming; lovemaps, boy and girl; rehearsal play; where do babies come from; sleeping and bathing] [tutorial 2] homosexuality, heterosexuality and gender-identity/role [prenatal hormonalization, postnatal gender differentiation, brain schemas] [tutorial 3] birth defect of the sex organs [neonatal sex announcement, telling others, parental sex life and genetic counseling, the child's sexuality, childhood counseling, teenage counseling, parable technique, chromosomal and gonadal sex, compliance, universals of the agenda] [tutorial 4] nosocomial neglect and abuse in pediatric sexology [corporal abuse and sexual abuse, genital exposure, the sexual reformation and counterreformation, the doctrine of wrong touching, juvenile sexual rehearsal play, lovemaps, lovemap disorder in a case of congenital heart disease, lovemap disorder in a case of female hermaphroditism with congenital virilizing adrenal hyperplasia (CVAH)] (PsycINFO Database Record (c) 2016 APA, all rights reserved)},
isbn={0-444-81262-8 (Hardcover)}

@Inbook{Utens2015,
author={Utens, Elisabeth M.
and Levert, Eveline},
title={Psychological aspects in children and adolescents with congenital heart disease and their parents.},
bookTitle={Clinical psychology and congenital heart disease: Lifelong psychological aspects and interventions.},
year={2015},
publisher={Springer-Verlag Publishing},
address={Utens, Elisabeth M.: Department of Child and Adolescent Psychiatry, Erasmus Medical Center, Sophia Children's Hospital, 2060, Rotterdam, Netherlands, 3000 CB, e.utens@erasmusmc.nl},
pages={35-52},
keywords={*Client Characteristics; *Clinical Psychology; *Congenital Disorders; *Heart Disorders; *Parent Child Relations; Behavior Problems; Cognitive Ability; Motor Skills; Parents; Quality of Life; Social Functioning; Social Skills},
abstract={Children with congenital heart disease (ConHD) constitute a heterogeneous group, in which mild and severe cardiac diagnoses are represented. The dramatic improvements of medical and surgical treatment for children with ConHD have led to increased survival rates during the last

decades, also for children with the most severe conditions. Whereas in former days, about 85 \% of the children born with ConHD died, nowadays about 90 \% of the children with ConHD survive into adulthood. Due to this hugely improved survival rate, the focus has shifted from mortality to morbidity. Children with ConHD may have cardiac residua and .sequelae after surgical or interventional treatment, and lifelong medical follow-up is recommended. With the focus on morbidity and ConHD now being considered more as a chronic condition, this has put forward the question: "What is the psychosocial impact of living with a congenital heart disease, both for parents and their children and the parent-child interaction?" The aim of this chapter is to give an overall outline of psychosocial aspects and problem areas, related to living with a ConHD, both for parents and their children, for different life phases and the parent-child interaction. Following the chronological order from birth to adulthood, we will discuss psychosocial aspects and problems for: (1) Parents of children with ConHD (2) The parent-child relationship (3) Preschoolers and school-age children with ConHD (4) Adolescents and transition-related topics. In these four parts, the following aspects related to living with a ConHD will be addressed: (a) emotional/behavioral problems, (b) intellectual and academic functioning, (c) quality of life, (d) motoric functioning, and (e) social functioning. Finally, a clinical case will be presented. This chapter does not aim to give a complete review, but rather focus on psychological areas, where a psychologist could provide help and counseling attuned to the needs of the individual child and adolescent with ConHD and the parents. Each of the four parts will be ended by conclusions and practical implications. (PsycInfo Database Record (c) 2020 APA, all rights reserved)),

isbn={978-88-470-5698-5 (Hardcover); 978-88-470-5699-2 (Digital (undefined format))},

doi={10.1007/978-88-470-5699-2_3},

url={https://doi.org/10.1007/978-88-470-5699-2_3}

@Inbook{Hampton2020,

author={Hampton, Lyla E.

and Demianczyk, Abigail C.

and Hoffman, Casey},

title={Cardiovascular disorders: Congenital heart disease and pediatric acquired cardiovascular disease.},

bookTitle={Pediatric health conditions in schools: A clinician's guide for working with children, families, and educators.},

year={2020},

publisher={Oxford University Press},

address={New York, NY, US},

pages={309-326},

keywords={*Adolescent Development; *Cardiovascular Disorders; *Congenital Disorders; *Early Childhood Development; *Quality of Life; Comorbidity; Evidence Based Practice; Life Span; Resilience (Psychological); Social Functioning},

abstract={Congenital heart disease (CHD) is one of the most common birth defects in the United States, affecting 1\% of births per year. Despite improved survival, individuals with CHD remain at high risk for neurological, cognitive, and psychosocial challenges that affect quality of life across the lifespan. CHD can be detected prenatally by genetic testing, obstetric ultrasound, or fetal echocardiogram. Early detection is encouraged to allow for genetic and family counseling as well as management of high-risk pregnancies. This chapter discusses the types of heart conditions and school-related concerns. Children with moderate to severe forms of CHD are at risk for neurobehavioral and school-related deficits. The deficits can be prevalent but subtle across early childhood to adolescence. The chapter discusses the comorbid neurodevelopmental diagnoses such as social deficits; and attention deficit hyperactivity disorder. It provides considerations for assessment of cognitive and neurodevelopmental conditions. The chapter discusses psychological adjustment and

health-related quality of life outcomes such as medical treatments and adjustment; developmental course of adjustment difficulties; family factors and adjustment. It discusses the risk and resiliency factors such as neurodevelopmental outcomes; and psychological maladjustment. The chapter explains evidence-based interventions; cultural considerations; and legal and policy issues. (PsycInfo Database Record (c) 2022 APA, all rights reserved)},
isbn={9780190687281 (Hardcover)},
doi={10.1093/med-psych/9780190687281.003.0017},
url={https://doi.org/10.1093/med-psych/9780190687281.003.0017}

@Article{Perelini2015,
author={Perelini, Fiona
and Blair, Nikki
and Wilson, Nigel
and Farrell, Alan
and Aitken, Andrew},
title={Family acceptability of school-based echocardiographic screening for rheumatic heart disease in a high-risk population in New Zealand.},
journal={Journal of Paediatrics and Child Health},
year={2015},
publisher={Wiley-Blackwell Publishing Ltd.},
address={Blair, Nikki: Department of Paediatrics, Wellington Hospital, Private Bag 7902, Wellington South, Wellington, New Zealand, 6021, nikki.blair@ccdhb.org.nz},
volume={51},
pages={682-688},
keywords={*At Risk Populations; *Cardiography; *Health Screening; *Parental Attitudes; *Rheumatic Fever; Disease Screening; Heart Disorders; Parent School Relationship; Pediatrics},
abstract={Aim: Echocardiographic screening for rheumatic heart disease has been piloted in high-risk areas in New Zealand and internationally, and fulfils most of the criteria for a targeted screening programme. The question of acceptability of rheumatic heart disease screening has not been assessed, and the aim of our study was to assess parental acceptability of a school-based echocardiographic screening programme in a high-risk population in New Zealand. Methods: A post-screening questionnaire was developed to survey parents of children who underwent echocardiographic screening. The families of 34 children with abnormal scan results and a sample of 80 children with normal scan results were surveyed by phone within 4 months of screening. Results: Positive results were seen in all survey questions in both normal and abnormal scan groups. All families were supportive of an ongoing screening programme. Of children with abnormal results, 62{\%} of their parents reported that they would treat their child differently; however, all responses were positive health-promoting outcomes. Conclusion: The study showed strong positive support for school-based echocardiographic screening by a community with high acute rheumatic fever incidence. The study did not detect any short-term negative effects in those with abnormal results. The survey result shows family and community support for the establishment of echocardiographic screening programmes in high acute rheumatic fever areas provided there is adequate infrastructural support. (PsycInfo Database Record (c) 2020 APA, all rights reserved)},
doi={10.1111/jpc.12829},
url={https://doi.org/10.1111/jpc.12829}

@Article{Ahmad2021,
author={Ahmad, Aaliya

and Fitzgerald-Butt, Sara M.

and Ware, Stephanie M.

and Ison, Hannah E.

and Elmore, Lindsey R.

and Helm, Benjamin M.},

title={Assessing genetic counselors' graduate school education and training in congenital heart defects.},

journal={Journal of Genetic Counseling},

year={2021},

publisher={Wiley-Blackwell Publishing Ltd.},

address={United Kingdom},

pages={No Pagination Specified-No Pagination Specified},

abstract={Genetic counselors are one of the many providers involved in caring for patients with congenital heart defects (CHDs); however, little is known about the cardiovascular genetics training they receive by their graduate programs. To explore the recalled education experiences regarding CHDs by practicing genetic counselors, we surveyed graduates of programs primarily accredited by the American Council on Genetic Counseling (ACGC) about their graduate training in this area, the depth of CHD-specific education they received, and whether CHDs are a substantial referral indication in their current practice. Genetic counselors were recruited from the National Society of Genetic Counselors and Twitter (n = 112), and participants reflecting multiple specialties and 35 graduate programs completed an online survey which included questions about fieldwork placements and lectures in cardiovascular genetics, exposure to classification schemes regarding cardiac embryology, and education in counseling strategies for CHDs and CHD-related topics during their graduate training. When asked whether CHDs are a substantial referral indication seen in their current practice, 55% (62/112) responded yes. Most participants (79% (88/112)) recalled receiving some education about CHDs, but 91% (80/88) reported receiving little to no education regarding embryologic classification of CHDs and how to apply classification schemes to their counseling. Both participating prenatal and pediatric GCs reported that CHDs can be a common referral indication, yet they reported receiving limited education on teratogens associated with CHDs, family screening recommendations, and recurrence risk counseling for CHDs. Based on participant responses, the majority of respondents reported receiving sufficient education on syndromes with CHDs which can be beneficial in specialties such as pediatrics. This exploratory study provides insight into opportunities to further support genetic counseling educational opportunities for CHDs. These findings suggest genetic counseling graduate programs could consider implementing education on CHD counseling strategies as a standardized component of the curriculum and that practicing genetic counselors could benefit from educational opportunities and resources with updated information on this topic. (PsycInfo Database Record (c) 2021 APA, all rights reserved)},

doi={10.1002/jgc4.1540},

url={https://doi.org/10.1002/jgc4.1540}

@Article{Deutch2020,

author={Deutch, Natalie

and Soo-Jin Lee, Sandra

and Char, Danton},

title={Translating genomic testing results for pediatric critical care: Opportunities for genetic counselors.},

journal={Journal of Genetic Counseling},

year={2020},

publisher={Wiley-Blackwell Publishing Ltd.},

address={Char, Danton: Department of Anesthesiology, Perioperative and Pain Management, Stanford University School of Medicine, Stanford University, Stanford, CA, US, dchar@stanford.edu},
volume={29},
pages={78-87},
keywords={*Genetic Counseling; *Genetics; *Health Personnel Attitudes; *Heart Disorders; Pediatrics},
abstract={Genomic sequencing (GS), such as whole genome and exome sequencing, is rapidly being integrated into pediatric critical care settings. Results are being used to make high impact decisions including declarations of futility, withdrawal of care, and rationing of scarce resources. In this qualitative study, we conducted interviews with clinicians involved in the care of critically ill children with congenital heart disease (CHD) to investigate their views on implementation of GS into clinical practice. Interviews were transcribed and inductively analyzed for major themes using grounded theory and thematic analysis. Three major themes emerged surrounding the use of genomic information in the high-stakes, time pressured decision making that characterizes clinical care of critically ill children with CHD: (a) that clinicians felt they did not have sufficient training to accurately assess genetic results despite pressure to incorporate results into clinical decisions; (b), that they desire knowledge support from genetic specialists, such as genetic counselors, who both understand the critical care context and are available within the time constraints of critical care clinical pressures; and (c), that clinicians feel a pressing need for increased genetics education to be able to safely and appropriately incorporate GS results into clinical decisions Our data suggest that genetics specialists may need a stronger presence in the pediatric critical care setting. (PsyInfo Database Record (c) 2022 APA, all rights reserved)},
doi={10.1002/jgc4.1182},
url={https://doi.org/10.1002/jgc4.1182}

@Article{Rombeek2020,
author={Rombeek, Meghan
and De Jesus, Stefanie
and Prapavessis, Harry
and Dempsey, Adam A.
and Fraser, Douglas
and Welisch, Eva
and Altamirano-Diaz, Luis
and Norozi, Kambiz},
title={Improving remote lifestyle intervention studies in children: Participant and caregiver feedback of the smart heart study.},
journal={Patient Education and Counseling},
year={2020},
publisher={Elsevier Science},
address={Netherlands},
pages={No Pagination Specified-No Pagination Specified},
abstract={OBJECTIVES: We sought to describe and analyze the quantitative and qualitative feedback obtained from participants and their caregivers of the Smart Heart study, a successful 12-month lifestyle intervention for children with overweight or obesity and congenital heart disease that provided remote lifestyle counseling, to improve future lifestyle interventions in children. METHODS: Thirty-six participants and caregivers were polled using a standard program evaluation questionnaire at the end of the intervention. Feedback was compiled into themes, and facilitators and barriers to program success were identified. RESULTS: There was a high level of satisfaction with the intervention and staff interactions as well as a low perceived burden of participation. There were also specific concerns, including mixed impressions regarding technology usage and a less impressive indication of actual

impactful behavior change. CONCLUSIONS: The study identified five themes, and corresponding facilitators and barriers to participant compliance, from the Smart Heart intervention feedback and offered suggestions for improving future lifestyle behavioral intervention study designs in children. PRACTICE IMPLICATIONS: Remote smartphone counseling is effective and efficient. It is recommended that the counseling messages are specific, the counseling schedule is patient-centric, patient burden is limited, methods with immediate patient feedback are used and family is included when feasible. (PsycInfo Database Record (c) 2020 APA, all rights reserved)},
doi={10.1016/j.pec.2020.02.016},
url={https://doi.org/10.1016/j.pec.2020.02.016}

@Article{Simko2005,
author={Simko, Lynn Coletta
and McGinnis, Kathleen A.},
title={What is the perceived quality of life of adults with congenital heart disease and does it differ by anomaly?},
journal={Journal of Cardiovascular Nursing},
year={2005},
publisher={Lippincott Williams {&} Wilkins},
address={Simko, Lynn Coletta: School of Nursing, Duquesne University, 600 Forbes Ave, Pittsburgh, PA, US, 15282, Simko@duq.edu},
volume={20},
pages={206-214},
keywords={*Client Attitudes; *Heart Disorders; Quality of Life},
abstract={Adults with congenital heart disease (CHD) represent a growing population of patients thanks to the medical and surgical advances which enable at least 85{\%} of children to survive to adulthood. These advances may create quality-of-life (QoL) issues not previously considered. The purpose of this cross-sectional study of 124 adults with CHD was to describe their QoL as a basis for providing appropriate information, counseling, and anticipatory guidance. Thirteen patients had single ventricle physiology (SVP), 43 had cyanotic lesions with 2 ventricle repairs, and 68 had acyanotic CHD. On the basis of Sickness Impact Profile (SIP) scores, individuals with SVP had worse QoL than did those with cyanotic lesions (with 2 ventricle repairs) and acyanotic anomalies (SIP = 9.98 vs 4.61 and 3.76). SIP scores were statistically significantly different between those with SVP and those with acyanotic anomalies (P = .02). For all groups, the areas of life most affected were work and sleep and rest. Participants with SVP saw themselves as having the poorest QoL. (PsycINFO Database Record (c) 2020 APA, all rights reserved)},
doi={10.1097/00005082-200505000-00013},
url={https://doi.org/10.1097/00005082-200505000-00013}

@PhdThesis{Smorra2020,
author={Smorra, Corinne},
title={Social workers' experiences with interventions for clients with congenital heart disease.},
year={2020},
publisher={ProQuest Information {&} Learning},
address={US},
volume={81},
pages={No Pagination Specified-No Pagination Specified},
keywords={*Awareness; *Heart Disorders; *Mental Health; *Social Workers; *Workplace Intervention; Clients},

abstract={Many adults born with congenital heart disease (CHD) face long-term psychosocial issues related the disease. The purpose of this project was to better understand social workers' experiences with interventions for clients who have CHD and experience psychosocial problems. Bronfenbrenner's e},
issn={0419-4217(Print)}

APÊNDICE H - RESULTADOS DE BUSCA NO GOOGLE ACADÊMICO

@Article{Keelan2022,
author={Keelan, Jenna A.
and Moon Grady, Anita J.
and Arya, Bhawna
and Donofrio, Mary T.
and Schidlow, David N.
and Tacy, Theresa A.
and Stern, Kenan W.D.
and Geiger, Miwa K.},
title={Current State of Fetal Heart Disease Counseling and Training: Room for Improvement?:
Endorsed by the Fetal Heart Society},
journal={Pediatric Cardiology},
year={2022},
month={Oct},
publisher={Springer},
keywords={Fellowship education; Fetal counseling; Fetal echocardiography},
abstract={We sought to describe the fellowship experiences and current practice habits of pediatric cardiologists who counsel patients with fetal heart disease (FHD) and to identify fellowship experiences related to FHD counseling perceived as valuable by respondents as well as opportunities for improvement. A cross-sectional survey of attending pediatric cardiologists who care for patients with FHD was performed. The respondents' demographics, fellowship experiences related to FHD counseling, reflections on fellowship training, and current practice habits were collected. The Fetal Heart Society endorsed this survey. There were 164 survey responses. 56{\%} of respondents did not have 4th-year subspecialty training in fetal cardiology. Observing and performing FHD counseling were the most commonly used methods of training, with the highest perceived effectiveness. The number of counseling sessions observed and performed correlated moderately with confidence in FHD counseling skills at fellowship graduation. Extracardiac pathology and neurodevelopment were the least frequently addressed topics in fellowship training and in current practice. Fewer than 50{\%} of respondents received formal education and feedback in counseling techniques during fellowship training. A significant proportion of practicing pediatric cardiologists provide FHD counseling with only standard categorical training. This highlights the potential importance of expanding FHD counseling education into categorical fellowship curricula. We suggest increasing opportunities for fellows to perform FHD counseling and receive feedback as this is a valued and beneficial experience during training. A formalized curriculum including extracardiac pathology and neurodevelopment and the use of evidence-based workshops in counseling techniques may address identified gaps in fellowship education.},
doi={10.1007/S00246-022-02882-4},
url={https://doi.org/10.1007/S00246-022-02882-4}}

@Article{Słodki2022,
author={Słodki, Maciej
and Copel, Joshua A.
and Rizzo, Giuseppe
and Araujo Junior, Edward
and Axt-Flidner, Roland
and Abuhamad, Alfred}

and Simpson, Lynn L.
and Lee, Wesley
and DeVore, Gregory
and Bahado-Singh, Ray
and Preis, Krzysztof
and Respondek-Liberska, Maria},
title={Fetal Cardiology: Is It Time to Establish a Separate Independent Medicine Subspecialty?},
journal={Pediatric Cardiology},
year={2022},
month={Oct},
publisher={Springer},
doi={10.1007/S00246-022-02936-7},
url={https://doi.org/10.1007/S00246-022-02936-7}

@Article{Ison2022,
author={Ison, Hannah E.
and Griffin, Emily L.
and {Parrott}
and {Ashley}
and Shikany, Amy R.
and {Meyers}
and {Lindsay}
and {Matthew}
and Thomas, J.
and Syverson, Erin
and Demo, Erin M.
and Fitzgerald, Kristi K.
and Fitzgerald-Butt, Sara
and {Katie}
and Ziegler, L.
and {Allison}
and Schartman, F.
and {Kristyne}
and Stone, M.
and {Benjamin}
and Helm, M.},
title={Genetic counseling for congenital heart disease--Practice resource of the national society of genetic counselors},
journal={Wiley Online Library},
year={2022},
month={Feb},
publisher={John Wiley and Sons Inc},
volume={31},
number={1},
pages={31-31},
keywords={complex disease; congenital heart disease; genetic counseling; genetic testing; practice resource},
abstract={Congenital heart disease (CHD) is an indication which spans multiple specialties across various genetic counseling practices. This practice resource aims to provide guidance on key considerations when approaching counseling for this particular indication while recognizing the rapidly

changing landscape of knowledge within this domain. This resource was developed with consensus from a diverse group of certified genetic counselors utilizing literature relevant for CHD genetic counseling practice and is aimed at supporting genetic counselors who encounter this indication in their practice both pre-and postnatally. **K E Y W O R D S** complex disease, congenital heart disease, genetic counseling, genetic testing, practice resource 1 | **I N T R O D U C T I O N** Congenital heart disease/defects (CHD/CHDs) constitute a heter-ogenous group of cardiovascular malformations and represent the most common birth defects in humans. These malformations have a birth incidence of 0.8{\%}-1{\%}, with an estimated global prevalence of approximately 1{\%}-2{\%} due to improved diagnosis, surgical interventions , and survival in the last three decades (Pierpont et al., 2018). The prevalence of adults with CHDs is now greater than that of children , with approximately 90{\%} of individuals surviving into adulthood},

doi={10.1002/jgc4.1498},

url={https://onlinelibrary.wiley.com/doi/abs/10.1002/jgc4.1498},

url={https://doi.org/10.1002/jgc4.1498}

@Article{DeBoever2022,

author={De Boever, Patrick

and Saguner, Ardan Muammer

and Kovacevic, Alexander

and Wacker-Gussmann, Annette

and B{"a}r, Stefan

and Els{"a}sser, Michael

and Motlagh, Aida Mohammadi

and Ostermayer, Eva

and Oberhoffer-Fritz, Renate

and Ewert, Peter

and Gorenflo, Matthias

and Starystach, Sebastian},

title={Fetal Cardiac Services during the COVID-19 Pandemic: How Does It Affect Parental Counseling?},

journal={mdpi.com},

year={2022},

keywords={fetal cardiology; parental counseling; parental needs; social science},

abstract={Citation: Kovacevic, A.; Wacker-Gussmann, A.; B{"a}r, S.; Els{"a}sser, M.; Mohammadi Motlagh, A.; Ostermayer, E.; Oberhoffer-Fritz, R.; Ewert, P.; Gorenflo, M.; Starystach, S. Parents' Perspectives on Abstract: After diagnosis of congenital heart disease (CHD) in the fetus, effective counseling is considered mandatory. We sought to investigate which factors, including parental social variables, significantly affect counseling outcome. A total of n = 226 parents were recruited prospectively from four national tertiary medical care centers. A validated questionnaire was used to measure counseling success and the effects of modifiers. Multiple linear regression was used to assess the data. Parental perception of interpersonal support by the physician ($\beta = 0.616$ ***, p = 0.000), counseling in easy-to-understand terms ($\beta = 0.249$ ***, p = 0.000), and a short period of time between suspicion of fetal CHD, seeing a specialist and subsequent counseling ($\beta = 0.135$ **, p = 0.006) significantly improve "overall counseling success". Additional modifiers (e.g., parental native language and age) influence certain subdimensions of counseling such as "trust in medical staff" (language effect: $\beta = 0.131$ *, p = 0.011) or "perceived situational control" (age effect: $\beta = 0.166$ *, p = 0.010). This study identifies independent factors that significantly affect counseling outcome overall and its subdimensions. In combination with existing recommendations our findings may contribute to more effective parental counseling. We further conclude that implementing communication skills training for specialists should be considered essential.},

doi={10.3390/jcm11010278},
url={https://www.mdpi.com/2077-0383/10/15/3423},
url={https://doi.org/10.3390/jcm11010278}

@Article{Goncalves2022,
author={Goncalves, Luis F.
and Lindblade, Christopher L.
and Cornejo, Patricia
and Patel, Mittun C.
and McLaughlin, Ericka Scheller
and Bardo, Dianna M.E.},
title={Contribution of fetal magnetic resonance imaging in fetuses with congenital heart disease},
journal={Pediatric Radiology},
year={2022},
month={Mar},
publisher={Springer Science and Business Media Deutschland GmbH},
volume={52},
number={3},
pages={513-526},
keywords={Anomalies; Brain; Congenital heart disease; Echocardiography; Fetus; Heart; Magnetic resonance imaging; Prenatal ultrasound; Pulmonary lymphangiectasia},
abstract={Background: Increasing evidence supports an association among congenital heart disease (CHD), structural brain lesions on neuroimaging, and increased risk of neurodevelopmental delay and other structural anomalies. Fetal MRI has been found to be effective in demonstrating fetal structural and developmental abnormalities. Objective: To determine the contribution of fetal MRI to identifying cardiovascular and non-cardiovascular anomalies in fetuses with CHD compared to prenatal US and fetal echocardiography. Materials and methods: We performed a retrospective study of fetuses with CHD identified by fetal echocardiography. Exams were performed on 1.5-tesla (T) or 3-T magnets using a balanced turbo field echo sequence triggered by an external electrocardiogram simulator with a fixed heart rate of 140 beats per minute (bpm). Fetal echocardiography was performed by pediatric cardiologists and detailed obstetrical US by maternal–fetal medicine specialists prior to referral to MRI. We compared the sensitivity of fetal MRI and fetal echocardiography for the diagnosis of cardiovascular anomalies, as well as the sensitivity of fetal MRI and referral US for the diagnosis of non-cardiac anomalies. We performed statistical analysis using the McNemar test. Results: We identified 121 anomalies in 31 fetuses. Of these, 73 (60.3{\%}) were cardiovascular and 48 (39.7{\%}) involved other organ systems. Fetal echocardiography was more sensitive for diagnosing cardiovascular anomalies compared to fetal MRI, but the difference was not statistically significant (85.9{\%}, 95{\%} confidence interval [CI] 77.8--94.0{\%} vs. 77.5{\%}, 95{\%} CI 67.7--87.2{\%}, respectively; McNemar test 2.29; P=0.13). The sensitivity of fetal MRI was higher for diagnosing extracardiac anomalies when compared to referral US (84.1{\%}, 95{\%} CI 73.3--94.9{\%} vs. 31.8{\%}, 95{\%} CI 18.1--45.6{\%}, respectively; McNemar test 12.9; P<0.001). The additional information provided by fetal MRI changed prognosis, counseling or management for 10/31 fetuses (32.2{\%}), all in the group of 19 fetuses with anomalies in other organs and systems besides CHD. Conclusion: Fetal MRI performed in a population of fetuses with CHD provided additional information that altered prognosis, counseling or management in approximately one-third of the fetuses, mainly by identifying previously unknown anomalies in other organs and systems.},
doi={10.1007/S00247-021-05234-1},
url={https://doi.org/10.1007/S00247-021-05234-1}

@Article{Kovacevic2022,
author={Kovacevic, A.
and Wacker-Gu{\ss}mann, A.
and B{\\"a}r, S.
and Els{\\"a}sser, M.},
title={Parents' perspectives on counseling for fetal heart disease},
year={2022},
url={https://katalog.ub.uni-heidelberg.de/titel/68863865}

@Article{Russo2022,
author={Russo, M. G.
and Fratta, F.
and Giudicepietro, A.
and Morelli, C.},
title={The Impact of Fetal Echocardiography on the Prognosis of Congenital Heart Disease},
year={2022},
url={https://www.intechopen.com/online-first/81805}

@Article{Malho2022,
author={Malho, Andr{\\"e} Souza
and Bravo-Valenzuela, Nathalie Jeanne
and Ximenes, Renato
and Peixoto, Alberto Borges
and J{\\"u}nior, Edward Araujo},
title={Antenatal diagnosis of congenital heart disease by 3D ultrasonography using spatiotemporal image correlation with HDlive Flow and HDlive Flow silhouette rendering modes},
journal={Ultrasonography},
year={2022},
month={Jul},
publisher={Korean Society of Ultrasound in Medicine},
volume={41},
number={3},
pages={578-596},
keywords={Congenital heart disease; Fetal heart; HDlive; Spatiotemporal image correlation; Three-dimensional ultrasound},
abstract={This pictorial review describes the assessment of a great variety of types of congenital heart disease by three-dimensional ultrasonography with spatiotemporal image correlation using HDlive and the HDlive Flow silhouette rendering mode. These technologies provide fetal heart surface patterns by using a fixed virtual light source that propagates into the tissues, permitting a detailed reconstruction of the heart structures. In this scenario, ultrasound operators can freely select a better light source position to enhance the anatomical details of the fetal heart. HDlive and the HDlive Flow silhouette rendering mode improve depth perception and the resolution of anatomic cardiac details and blood vessel walls compared to standard two-dimensional ultrasonography.}

@Article{Simpson2022,

author={Simpson, John
and Hornberger, Lisa K.},
title={What Does Fetal Echocardiography Add Beyond the Anomaly Scan?},
journal={Circulation: Cardiovascular Imaging},
year={2022},
month={Apr},
publisher={Lippincott Williams and Wilkins},
volume={15},
number={4},
pages={e014168-e014168},
keywords={Editorials; fetal heart; heart defects, congenital; prenatal diagnosis; prognosis},
doi={10.1161/CIRCIMAGING.122.014168},
url={https://doi.org/10.1161/CIRCIMAGING.122.014168}

@Article{Zloto2022,
author={Zloto, Keren
and Hochberg, Alyssa
and Tenenbaum-Gavish, Kinneret
and Berezowsky, Alexandra
and Barbash-Hazan, Shiri
and Bardin, Ron
and Hadar, Eran
and Shmueli, Anat},
title={Fetal congenital heart disease - mode of delivery and obstetrical complications},
journal={BMC Pregnancy and Childbirth},
year={2022},
month={Dec},
publisher={BioMed Central Ltd},
volume={22},
number={1},
keywords={Congenital heart diseases; Non-reassuring fetal heart rate; Operative vaginal delivery},
abstract={Background: The optimal mode of delivery in cases of fetal congenital heart disease (CHD) is not established. The few relevant studies did not address operative vaginal delivery. The aim of this study was to assess the impact of fetal CHD on mode of delivery during a trial of labor, and to secondarily describe some obstetric complications. Methods: The database of a tertiary medical center was searched for women who gave birth to a singleton, liveborn neonate in 2015--2018. Mode of delivery was compared between women carrying a fetus with known CHD and women with a healthy fetus matched 1:5 for maternal age, parity, body mass index, and gestational age. Results: The cohort included 616 women, 105 in the CHD group and 511 in the control group. The rate of operative vaginal delivery was significantly higher in the CHD group (18.09% vs 9.78%, OR 2.03, 95% CI 1.13--3.63, p = 0.01); the difference remained significant after adjustment for nulliparity and gestational age at delivery (aOR 2.58, 95% CI 1.36--4.9, p < 0.01). There was no difference between the CHD and control group in rate of intrapartum cesarean delivery (9.52% vs 10.76%, respectively, OR 0.97, 95% CI 0.47--1.98, p = 0.93). The most common indication for operative vaginal delivery was non-reassuring fetal heart rate (78.94% vs 64%, respectively). Median birth weight percentile was significantly lower in the CHD group (45th vs 53rd percentile, p = 0.04). Conclusions: Our findings suggest that operative vaginal delivery, performed mostly because of non-reassuring fetal heart rate, is more common in pregnancies complicated by a prenatal diagnosis of CHD than non-anomalous pregnancies.},
doi={10.1186/S12884-022-04910-W},

url={<https://doi.org/10.1186/S12884-022-04910-W>}

@Article{Karmegaraj2022,
author={Karmegaraj, Balaganesh
and Udhayakumar, Vani
and Selvan, Gigi},
title={First Trimester Prenatal Diagnosis of a Conotruncal Anomaly Using Spatiotemporal Image
Correlation Imaging Confirmed by Conventional Autopsy},
journal={Fetal and Pediatric Pathology},
year={2022},
publisher={Taylor and Francis Ltd.},
volume={41},
number={2},
pages={346-350},
keywords={Fetus; Truncus arteriosus; conventional autopsy; first trimester; spatiotemporal image
correlation imaging},
abstract={Background Fetal echocardiography continues to be the first line investigation for detecting
congenital heart diseases (CHD). As accurate and complete diagnosis of complex heart disease is
often difficult in the first trimester due to small size of the fetal heart, confirmation/expanded
description by fetopsy provides the best information for accurate counseling for future pregnancies.
Although non invasive fetal autopsy alternatives have been investigated with favorable results,
conventional autopsy remains the gold standard procedure used to confirm the fetal abnormalities.
Case report: We describe a conotruncal anomaly diagnosed at 12 weeks gestation using
spatiotemporal image. The fetopsy confirmed the diagnosis of Type I Truncus arteriosus. Conclusion:
Four-dimensional STIC imaging provides incremental benefits in evaluation of fetal cardiac anomalies,
and confirmation by autopsy findings allows further refinement of the diagnosis.},
doi={10.1080/15513815.2020.1806419},
url={<https://doi.org/10.1080/15513815.2020.1806419>}

@Article{Gowda2022,
author={Gowda, M.
and Thiagarajan, M.
and {ldots}, S. Satheesh -. The Journal of
and 2022, undefined},
title={Prenatal grading of fetal congenital heart disease and its influence on decision making during
pregnancy and postnatal period: a prospective study},
journal={Taylor {&} Francis},
year={2022},
publisher={Taylor and Francis Ltd.},
volume={35},
number={16},
pages={3158-3166},
keywords={Congenital cardiac defects; prenatal grading of CHDs; prenatal prognostication of CHDs;
prognosticating CHDs},
abstract={Background: Congenital heart defects(CHDs) are an important cause of neonatal mortality
and morbidity. With advances in diagnosis and treatment, many defects are now amenable to
correction. There is a need for individualized approach to prenatally detected lesions to predict the
likely prognosis. Assigning them into risk category helps in prenatal counseling, decision making,

referrals and formulation of management plan to improve the outcome. Objective: To grade the fetal CHDs according to severity and study its usefulness in decision making. Methods: A prospective study at a tertiary care institute between 2016 and 18, including pregnant women with antenatal diagnosis of fetal CHD. Detailed fetal echocardiography was followed by classification of lesions into four risk categories using modified grading system: (A) extremely high risk; (B) high risk (C) moderate risk (D) low risk. Appropriate counseling was provided to facilitate decision making and further management. The grading was reviewed and revised again postpartum/post-mortem for correlation. Results: Of the total 137 cases, almost half (45.53%) were Category B, while Category D, C and A had 24.1%, 20.4% and 10.2% of cases respectively. The mean gestation age at diagnosis was 26.5 weeks. Termination of pregnancy was done in 21 cases, mostly in Category B (71.4%) and of the 116 continued pregnancies, there were 16 intrauterine deaths. Prenatal and postnatal findings were available in 109 cases and kappa analysis for agreement between antenatal and postnatal grading showed good agreement (0.82). Conclusion: Prenatal grading of congenital heart disease is a reliable, structured and simplified tool that can be used for providing counseling and facilitate decision making.},

doi={10.1080/14767058.2020.1814245},

url={https://www.tandfonline.com/doi/abs/10.1080/14767058.2020.1814245},

url={https://doi.org/10.1080/14767058.2020.1814245}

@Article{Steinweg2022,

author={Steinweg, J. K.

and Roberts, T. A.

and Van Poppel, M. P.

and Lloyd, D.

and Razavi, R.

and Hajnal, J. V.

and Pushparajah, K.},

title={Fetal Blood Flow Assessment Using Motion-Robust Whole Heart 4D Cine MRI},

journal={The 54th Annual Meeting of the German Society for Pediatric Cardiology (DGPK)},

year={2022},

month={Jan},

publisher={Georg Thieme Verlag KG},

volume={70},

doi={10.1055/S-0042-1743024},

url={https://doi.org/10.1055/S-0042-1743024}

@Article{Ruan2022,

author={Ruan, Yanping

and Xie, Zan

and Yantai, Aliated

and Liu, Xiaowei},

title={Associated Maternal And Fetal Factors Of Fetal Congenital Heart Diseases Diagnosed By Fetal Echocardiography},

year={2022},

keywords={Congenital heart diseases; Factor; Odd ratio; fetal},

abstract={Objective Current studies have suggested that fetal congenital heart diseases (CHDs) are caused by various factors. However, few data is available with respect to the aspect in China. This study aimed to detect associated maternal and fetal factors of fetal CHD in a large sample in China.

Study Design: Pregnant women who underwent fetal echocardiography (N = 5024) were recruited in our hospital between May 2018 and September 2019. Of these, 875 pregnant women had fetuses with CHD. The maternal sociodemographic and lifestyle characteristics and some fetal factors were obtained. We used forward stepwise logistic regression analysis to assess the associations of fetal CHD with various factors. Results Among the fetal CHD group (N = 875), critical CHDs account for 27%, of which Tetralogy of Fallot is the most (7.1%), followed by coarctation of aorta (4.0%), Double-outlet right ventricle (2.9%). The forward stepwise logistic regression models revealed that gravidity (OR = 1.32, 95%CI 1.21-1.43, P = 0.000), upper respiratory tract infection during early pregnancy (OR = 1.30, 95%CI 1.04-1.63, P = 0.021), mental stress during early pregnancy (OR = 2.87, 95%CI 1.35-5.92, P = 0.006), single umbilical artery (OR = 2.46, 95%CI 1.24-4.85, P = 0.010), and parental smoking (OR = 1.20, 95%CI 1.00-1.45, P = 0.048) are positively associated with an increased risk of fetal CHD. Conclusion We identified two maternal factors and two fetal factors positively associated with fetal CHD. These findings suggest that it is important to strengthen healthcare and prenatal counseling for women with these factors.},
url={<https://www.researchsquare.com/article/rs-1408405/latest.pdf>}

@Article{Stephens2022,
author={Stephens, Elizabeth H.
and Dearani, Joseph A.
and Mauermann, William
and Bendel-Stenzel, Ellen M.
and Arendt, Katherine W.
and Rose, Carl H.
and Blau, Caitlin
and Crestanello, Juan
and Schaff, Hartzell},
title={Cardiac Surgery during Pregnancy},
journal={annalsthoracicsurgeryshortrep.org},
year={2022},
keywords={cardiac surgery; maternal-fetal medicine; pregnancy},
abstract={This is a PDF file of an article that has undergone enhancements after acceptance, such as the addition of a cover page and metadata, and formatting for readability, but it is not yet the definitive version of record. This version will undergo additional copyediting, typesetting and review before it is published in its final form, but we are providing this version to give early visibility of the article. Please note that, during the production process, errors may be discovered which could affect the content, and all legal disclaimers that apply to the journal pertain.},
doi={10.1016/j.atssr.2022.08.004},
url={[https://www.annalsthoracicsurgeryshortrep.org/article/S2772-9931\(22\)00012-2/abstract](https://www.annalsthoracicsurgeryshortrep.org/article/S2772-9931(22)00012-2/abstract)},
url={<https://doi.org/10.1016/j.atssr.2022.08.004>}

@Article{Maternal2022,
author={Maternal, Weihai
and Health, Child
and Xinhong, Hospital
and Maternal, Yang Weihai
and Health Hospital, Child},
title={Application of chromosomal microarray analysis in congenital heart disease},
year={2022},

keywords={chromosomal microarray analysis; copy number variation; fetal congenital heart disease; prenatal diagnosis; sNP-array technology; ventricular septal defect},
isbn={2986860699},
url={https://www.researchsquare.com/article/rs-1933660/latest.pdf}

@Article{Lynch2022,
author={Lynch, Tara A.
and Westen, Elizabeth
and Li, Dongmei
and Katzman, Philip J.
and Malshe, Amol
and Drennan, Kathryn},
title={Stillbirth in women with diabetes: a retrospective analysis of fetal autopsy reports},
journal={Journal of Maternal-Fetal and Neonatal Medicine},
year={2022},
publisher={Taylor and Francis Ltd.},
volume={35},
number={11},
pages={2091-2098},
keywords={Fetal hypertrophic cardiomyopathy; autopsy; diabetes; stillbirth},
abstract={Introduction: Diabetes in pregnancy is associated with an increased rate of stillbirth. There are a wide variety of factors that have been implicated including placental insufficiency, hypoxia, hyperinsulinemia and impaired cardiac function. Furthermore, there is evidence that diabetic pregnancies have an increased rate of fetal cardiomyopathy as compared to non-diabetic pregnancies. Prior studies have indicated that this association can also be an etiology for diabetic stillbirth. The purpose of this study was to determine if diabetic pregnancies have an increased risk of fetal cardiomyopathy identified on fetal autopsy as compared to non-diabetic women with a stillbirth in a cohort of pregnancies that had evaluation with a fetal autopsy. Materials and methods: Retrospective cohort study of women with a stillbirth who consented to fetal autopsy at an academic medical center from 2011 to 2017. Stillbirth was defined as an intrauterine fetal demise at ≥ 20 weeks' gestation. Women with diabetes defined as pre-gestational diabetes type 1, pre-gestational diabetes type 2, and gestational diabetes were compared to women without diabetes. Primary outcome was fetal cardiomyopathy. Other etiologies for stillbirth were also evaluated and classified according to the Stillbirth Collaborative Research Network (SCRN) initial causes of fetal death. Fisher exact test, χ^2 test, and Mann Whitney U tests were performed as appropriate, with $p < .05$ considered significant. Generalized linear models were performed for fetal organ weights controlling for gestational age of delivery, maternal chronic hypertension, delivery body mass index, and birthweight. Results: A total of 78 women elected to have fetal autopsy examinations during the study period. Of these, 75 had complete information available for review. A total of 60 women did not have diabetes and 15 women had diabetes. Of pregnancies complicated by diabetes, 11 had insulin dependent diabetes and 4 had non-insulin dependent diabetes. Fetal cardiomyopathy was diagnosed on autopsy for 7 (46.7%) of pregnancies with diabetes and 2 (3.3%) of pregnancies without diabetes (RR 14.00 [95% CI 3.23--60.65], $p < .001$). These associations were still significant even when analyzing only those pregnancies without fetal congenital heart disease (7 [46.7%] diabetic pregnancies with cardiomyopathy versus 1 [2.0%] nondiabetic pregnancy with cardiomyopathy, RR 23.80 [95% CI 3.17--178.46], $p < .001$). There was no difference between diabetic and non-diabetic pregnancies in regards to other causes for stillbirth. Stillbirths in pregnancies with diabetes also had larger fetal heart, liver, and adrenal weights on fetal autopsy. Conclusion: Women with diabetes have 14 times the risk of fetal cardiomyopathy identified at fetal autopsy as compared to women without diabetes. As the

prediction and prevention of diabetic stillbirth is limited, information on potential causes of stillbirth may help future research identify those pregnancies at the greatest risk for adverse outcome.},
doi={10.1080/14767058.2020.1779213},
url={https://doi.org/10.1080/14767058.2020.1779213}

@Article{Savla2022,
author={Savla, Jill J.
and Putt, Mary E.
and Huang, Jing
and Parry, Samuel
and Moldenhauer, Julie S.
and Reilly, Samantha
and Youman, Olivia
and Rychik, Jack
and Mercer-Rosa, Laura
and Gaynor, ;. J. William
and Kawut, Steven M.},
title={Impact of Maternal--Fetal Environment on Mortality in Children With Single Ventricle Heart Disease},
journal={Am Heart Assoc},
year={2022},
month={Jan},
publisher={American Heart Association Inc.},
volume={11},
number={2},
pages={20299-20299},
keywords={Congenital heart disease; Fetal development; Fetal programming; Hypoplastic left heart syndrome; Preeclampsia/pregnancy; Prenatal exposures; Stage 1 Norwood procedure},
abstract={BACKGROUND: Children with single ventricle heart disease have significant morbidity and mortality. The maternal-fetal environment (MFE) may adversely impact outcomes after neonatal cardiac surgery. We hypothesized that impaired MFE would be associated with an increased risk of death after stage 1 Norwood reconstruction.},
doi={10.1161/JAHA.120.020299},
url={https://www.ahajournals.org/doi/abs/10.1161/JAHA.120.020299},
url={https://doi.org/10.1161/JAHA.120.020299}}

@Article{Bauser-Heaton2022,
author={Bauser-Heaton, Holly
and Gil, Carmen J.
and Serpooshan, Vahid},
title={Bioengineering of Pediatric Cardiovascular Constructs: In Vitro Modeling of Congenital Heart Disease},
journal={Advanced Technologies in Cardiovascular Bioengineering},
year={2022},
publisher={Springer International Publishing},
pages={233-248},
doi={10.1007/978-3-030-86140-7_11},
url={https://doi.org/10.1007/978-3-030-86140-7_11}}

@Article{Li2022a,
author={Li, S.
and Zhe, L.},
title={A retrospective analysis of fetal heart defects in China: Investigating the influencing factors of pregnancy outcomes and evaluating the value of the},
year={2022},
url={<https://www.researchsquare.com/article/rs-1745471/latest.pdf>}

@Article{Pick2022,
author={Pick, Justin
and Silka, Michael J.
and Bar-Cohen, Yaniv
and Hill, Allison
and Shwayder, Mark
and Wood, John
and Pruetz, Jay D.},
title={Third Trimester Fetal Heart Rates in Antibody-Mediated Complete Heart Block Predict Need for Neonatal Pacemaker Placement},
journal={Pediatric Cardiology},
year={2022},
month={Feb},
publisher={Springer},
volume={43},
number={2},
pages={324-331},
keywords={Bradycardia; Congenital heart block; Fetal cardiology; Fetal echocardiography; Lupus antibodies; Pacemaker; Prenatal diagnosis; Prenatal ultrasound},
abstract={Congenital complete heart block (CCHB) affects 1 in 20,000 newborns. This study evaluates fetal and neonatal risk factors predictive of neonatal pacemaker placement in antibody-mediated complete heart block. The Children's Hospital Los Angeles institutional fetal, pacemaker, and medical record databases were queried for confirmed SSA/SSB cases of CCHB between January 2004 and July 2019. Cases excluded were those with a diagnosis beyond the neonatal period, diagnosis of a channelopathy, or if maternal antibody status was unknown. We recorded the gestational age (GA), birth weight (BW), fetal heart rates (FHRs) of the last echocardiogram before delivery, specific neonatal ECG and echocardiogram findings, age at pacemaker placement, and mortality. Of 43 neonates identified with CCHB, 27 had confirmed maternal antibody exposure. Variables associated with neonatal pacemaker implantation were FHRs < 50 bpm ($p = 0.005$), neonatal heart rates < 52 bpm ($p = 0.015$), and neonatal left ventricular fractional shortening (FS) percentages < 34% ($p = 0.03$). On multivariate analysis, FHR remained significant ($p = 0.03$) and demonstrated an increased risk of neonatal pacemaker placement by an odds ratio of 12.5 (95% CI 1.3–116, $p = 0.05$). The median GA at which the FHR was obtained was 34 weeks (IQR 26–35 weeks). Neonatal pacemaker placement was highly associated with a FHR < 50 bpm, neonatal HR < 52 bpm, and neonatal FS < 34%. FHRs at 34 weeks GA (IQR 26–35 weeks) correlated well with postnatal heart rates and were predictive of neonatal pacemaker placement.},
doi={10.1007/S00246-021-02723-W},
url={<https://doi.org/10.1007/S00246-021-02723-W>}

@Article{Herghelegiu2022,
author={Herghelegiu, Catalin Gabriel
and Duta, Simona Florentina
and Neacsu, Adrian
and Suciu, Nicolae
and Veduta, Alina},
title={Operator experience impact on the evaluation of still images of a first trimester cardiac
assessment protocol},
journal={Journal of Maternal-Fetal and Neonatal Medicine},
year={2022},
publisher={Taylor and Francis Ltd.},
volume={35},
number={10},
pages={1957-1961},
keywords={3 vessel view; 4 Chamber view; congenital heart disease; fetal cardiac screening; first
trimester},
abstract={Objective: Congenital heart disease (CHD) is the most common birth defect and represents
the leading cause for mortality and morbidity in infants and young adults. Early fetal echocardiography
is usually considered a highly specialized scan. The goal of this study is to evaluate the impact of
operator's experience in assessing still images of the 4-chamber view and 3-vessels view and to
evaluate the feasibility and the performance of a first trimester screening protocol for CHD. Methods:
An online questionnaire consisting of still images of the 4-camber view and 3-vessel view from 50
normal and abnormal cases was reviewed by an expert group made of seven obstetricians specialized
in fetal medicine and a nonexpert group made of 13 obstetricians that are certified in ultrasound. After
individually visualizing each image set made of the 4-chamber view and 3-vessel view, they had to
conclude if the case was normal or abnormal and what images were abnormal. Results: A total of 50
image sets of both normal and abnormal fetal hearts were examined by the 20 reviewers, resulting in
1000 evaluations. The expert group achieved a detection rate of 97.1{\%} with a false positive rate of
5.7{\%}. The nonexpert group achieved also a good detection rate of 91.3{\%} but with a much higher
false positive rate of 33.9{\%}. The most frequently missed CHD involved the great arteries and had a
normal 4-chamber view. In the majority of false positive cases the 3-vessel view was incorrectly
interpreted as abnormal. Conclusions: A screening protocol for CHD, based on the 4-chamber view
and 3-vessel view alone can offer a good detection rate for CHD with a small false positive rate, but
only if it is implemented by highly specialized sonographers.},
doi={10.1080/14767058.2020.1774873},
url={https://doi.org/10.1080/14767058.2020.1774873}}

@Article{Fesslova2022,
author={Fesslova, V.
and Medicine, PI Cavoretto -. Journal of Clinical
and 2022, undefined},
title={Recent Advances in the Diagnosis and Management of Congenital Heart Defects},
journal={mdpi.com},
year={2022},
doi={10.3390/jcm11195534},
url={https://www.mdpi.com/2077-0383/11/19/5534/pdf?version=1663763663},
url={https://doi.org/10.3390/jcm11195534}}

@Article{NasrEldin2022,
author={Nasr Eldin, Aya S.
and Elshwaikh, Shereef L.
and Elhefnawy, Suzan B.
and Elkholi, Dina G.},
title={Value of First and Early Second Trimester Fetal Echocardiography and Fetal Nuchal
Translucency Measurement in Prediction of Fetal Congenital Heart Diseases},
journal={eprints.asianrepository.com},
year={2022},
volume={34},
number={1},
pages={73-87},
keywords={Trimester fetal; congenital heart diseases; echocardiography; fetal nuchal translucency},
abstract={Background: Congenital heart anomalies have a significant effect on affected children's life
with up to 25:35{\%} mortality rate during pregnancy and the postnatal period, and 60{\%} of this
mortality occurs during the first year of life. Objectives: To evaluate the value of fetal echocardiography
and measuring of fetal nuchal translucency thickness in first and early second trimester in prediction of
fetal congenital heart disease. Methods: A prospective observational cohort study was conducted on
200 pregnant women attended to Tanta University Hospital, Department of Obstetrics and Gynecology
at outpatient clinic or inpatient, the study took from January 2019 to December 2020. Patients
underwent transabdominal fetal echocardiography at 10:16 weeks of gestation. transvaginal scan was
also performed when needed. The first step in fetal cardiac ultrasound is to evaluate the orientation of
the fetal laterality (presentation and lie). Establishing situs and atrial arrangement, Five short-axis
views of heart. First view is acquired in abdomen at level of stomach to identify situs. Results: predict
congenital heart disease (compared to results of first Echo), nuchal translucency had 83.33{\%}
sensitivity, 98.97{\%} specificity, 71.43{\%} PPV, 98.50{\%} NPV and 99.48{\%} accuracy.. Original
Research Article EIDin et al.; JAMMR, 34(1): 73-87, 2022; Article no.JAMMR.82452 74 predict
congenital heart disease (compared to fetal outcome), nuchal translucency had 83.33{\%}
sensitivity,98.97{\%} specificity, 71.43{\%} PPV, 99.48{\%}{\%} NPV and 98.50{\%} accuracy.
Conclusions: Fetal echocardiography and measuring of fetal nuchal translucency thickness in first and
early second trimester have good sensitivity in prediction and better specificity in exclusion of fetal
congenital heart diseases.},
doi={10.9734/JAMMR/2022/v34i131250},
url={http://eprints.asianrepository.com/id/eprint/2413/},
url={https://doi.org/10.9734/JAMMR/2022/v34i131250}}

@Article{Wacker-Gussmann2022,
author={Wacker-Gussmann, A.
and Medicine, R. Oberhoffer-Fritz -. Journal of Clinical
and 2022, undefined},
title={Cardiovascular Risk Factors in Childhood and Adolescence},
journal={mdpi.com},
year={2022},
volume={2022},
pages={1136-1136},
abstract={According to the World Health Organization (WHO), cardiovascular diseases (CVDs) are the
leading cause of death globally, taking an estimated amount of 17.9 million lives each year. CVD is
therefore a major public health problem contributing to the global burden of morbidity and mortality. It

affects people over their whole lifespan, especially the elderly. Nowadays, cardiovascular (CV) risk factors are dramatically increasing in children and adolescents due to sociodemographic changes. The ongoing COVID-19 pandemic situation even worsens such developments. Physical distancing, distant learning and virtual schooling, exposure to increased screen time, and less physical activity have all contributed to increased weight gain among youth. Weight gain and obesity are associated with increased risk for atherosclerosis and, consequently, CVD in all ages. During the Life Course, there is also an accumulation of different risk factors contributing to the development of CVD: in addition to biological and behavioral factors, environmental and psychosocial factors affect well-being and CV health. Moreover, there is the role of the whole family with particular socioeconomic and lifestyle factors, which also have to be considered. In addition to these growing risk factors for acquired CVD in youth, there is still congenital heart disease (CHD) as the most common birth defect worldwide, affecting nearly one percent of newborns each year. With the development of better surgical techniques, infant mortality rates have dropped dramatically. This means that there is a growing population of adults with CHD. Therefore, environmental and lifestyle factors also influence patients with CHD and other chronic diseases, but this is outside of the research focus. This Special Issue published in the Journal of Clinical Medicine aimed to collect high-quality studies that represent the most recent advancements in CV research of children. We focused on different aspects of cardiovascular risks for CVD in childhood and adolescence, from pregnancy and fetal counseling [1] over childhood after high-risk pregnancies [2] to young adults. According to the key topics of Pediatric Cardiology, contributions to CHD and car-diomyopathies [3,4] as well as acquired heart disease were edited. Lifestyle factors, nutrition and physical activity [5,6] partly resulting in CV risk factors, such as obesity [7,8], were reported. Bjelakovic et al. [8] published a Position Paper from Serbia, which reported on the CV risk assessment and clinical management of children and adolescents with heterozygous familial hypercholesterolemia, which certainly has a great impact on CVD. Multiple interrelated influences with exponentially growing adverse CV effects over the lifespan impede research and raise the question: at which key stage of the life course does the prevention of CV risks in youth achieve the highest benefit for health? It has to be considered that prevention strategies in youth will not only reach adulthood of the individual itself. Due to the upcoming reproductive age, it will also at least reach the next generation. High-risk pregnancies with the effect of fetal programming, overweight and obesity, arterial hypertension, dyslipidemia and lifestyle factors such as physical activity,},

doi={10.3390/jcm11041136},

url={https://www.mdpi.com/2077-0383/11/4/1136/htm},

url={https://doi.org/10.3390/jcm11041136}

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author={Limperopoulos, Catherine

and Wessel, David L.

and Du Plessis, Adre J.},

title={Understanding the Maternal-Fetal Environment and the Birth of Prenatal Pediatrics},

journal={Journal of the American Heart Association},

year={2022},

month={Jan},

publisher={American Heart Association Inc.},

volume={11},

number={2},

keywords={Editorials; birth weight; child; fetal brain; humans; pediatrics; pregnancy},

doi={10.1161/JAHA.121.023807},

url={https://doi.org/10.1161/JAHA.121.023807}

@Article{Cao2022,
author={Cao, Y.
and {Chau}
and K, M. H.
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and {Kwan}
and W, A. H.
and {Hui}
and A, S. Y.
and {Lam}
and H, Y.
and {Tan}
and T, T. Y.
and {Tse}
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and {L}
and {Leung}
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and {Dong}
and {Z}
and {Choy}
and W, K.
and Choy, K. W.
and Dong, Z.},
title={Exploring the diagnostic utility of genome sequencing for fetal congenital heart defects},
journal={Wiley Online Library},
year={2022},
month={Jun},
publisher={John Wiley and Sons Ltd},
volume={42},
number={7},
pages={862-872},
abstract={Objective: The diagnostic yield for congenital heart defects (CHD) with routine genetic testing is around 10{\%}-20{\%} when considering pathogenic CNVs or aneu-ploidies as positive findings. This is a pilot study to investigate the utility of genome sequencing (GS) for prenatal diagnosis of CHD. Methods: Genome sequencing (GS, 30X) was performed on 13 trios with CHD for which karyotyping and/or chromosomal microarray results were non-diagnostic. Results: Trio GS provided a diagnosis for 4/13 (30.8{\%}) fetuses with complex CHDs and other structural anomalies. Findings included pathogenic or likely pathogenic variants in DNAH5, COL4A1, PTPN11, and KRAS. Of the nine cases without a genetic etiology by GS, we had outcome follow-up data on eight. For five of them (60{\%}), the parents chose to keep the pregnancy. A balanced translocation [46,XX,t(14; 22)(q32.33; q13.31)mat] was detected in a trio with biallelic DNAH5 mutations, which together explained the recurrent fetal situs inversus and dextrocardia that was presumably due to de novo Phelan-McDermid syndrome. A secondary finding of a BRCA2 variant and carrier status of HBB, USH2A, HBA1/HBA2 were detected in the cohort. Conclusions: GS expands the diagnostic scope of mutation types over conventional testing, revealing the genetic etiology for fetal heart anomalies. Patients without a known genetic abnormality indicated by GS likely opted to keep pregnancy

especially if the heart defect could be surgically repaired. We provide evidence to support the application of GS for fetuses with CHD. Key points What's already known about this topic? ♦ The diagnostic yield of exome sequencing for fetuses with structural abnormalities after undiagnostic conventional testing is 20%-30%. The diagnostic yield of genome sequencing for fetal heart defects is not established. Y. Cao, M. H. K. Chau, and Y. Zheng are Equally contributed to this work. 862-Prenatal Diagnosis. 2022;42:862-872. wileyonlinelibrary.com/journal/pd}, doi={10.1002/pd.6151}, url={https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/pd.6151}, url={https://doi.org/10.1002/pd.6151}

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author={Akalin, M.
and Yalcin, Murat
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and Akalin, Emine Eda
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title={Positive effects of fetal echocardiography on maternal anxiety: a prospective study in a tertiary center in Turkey},
journal={Journal of Psychosomatic Obstetrics & Gynecology},
year={2022},
month={Sep},
pages={1-8},
doi={10.1080/0167482X.2022.2124911},
url={https://www.tandfonline.com/doi/full/10.1080/0167482X.2022.2124911},
url={https://doi.org/10.1080/0167482X.2022.2124911}}

@Article{Cai2022,
author={Cai, M.},
title={Genetic etiology and obstetric outcome analysis of fetal cystic hygroma in a single-center study},
year={2022},
url={https://www.researchsquare.com/article/rs-1467309/latest.pdf}}

@Article{Madazli2022,
author={Madazli, R.
and Davutoglu, E. Alcin
and Alpay, V. - Journal of Obstetrics
and Gynecology, 2022, undefined},
title={Perinatal outcomes of pregnancies with prenatally diagnosed foetal congenital heart disease},
journal={Taylor & Francis},
year={2022},
publisher={Taylor and Francis Ltd.},

volume={42},
number={5},
pages={1079-1084},
keywords={Congenital heart disease; perinatal outcomes; prenatal diagnosis},
abstract={We aimed to assess the types of prenatally diagnosed congenital heart disease (CHD) and their association with structural and chromosomal abnormalities and to evaluate the perinatal outcomes according to the type of the heart defect. We retrospectively reviewed 377 pregnancies with prenatally diagnosed CHD. The main outcome measure was to evaluate the pregnancy outcomes of CHD according to the type of the heart defect and associated structural or chromosomal abnormalities. Of 377 fetuses with major structural CHD, 214 (56.8\{\%\}) were isolated, 49 (13\{\%\}) had additional cardiac anomalies, 58 (15.4\{\%\}) had extracardiac malformations with normal karyotype and 56 (14.9\{\%\}) had chromosomal abnormalities. The most common chromosomal abnormality was trisomy 21 (55.4\{\%\}). Prenatal detection of CHD allows early workup to identify chromosomal abnormalities and detailed anatomic evaluation of extracardiac malformations. Prognostication of each heart defect at diagnosis and facilitating patients with isolated surgically correctable CHD for targeted postnatal care is essential.IMPACT STATEMENTWhat is already known on this subject? CHD is the most common structural anomaly and is strongly associated with chromosomal anomalies and genetic syndromes. What do the results of this study add? Survival of the prenatally diagnosed CHD depends on the type and severity of the condition and coexisting extracardiac structural or chromosomal abnormalities. What are the implications of these findings for clinical practice and/or further research? Prenatal detection of CHD allows early workup to identify chromosomal abnormalities, detailed anatomic evaluation of extracardiac malformations and time to refer the parents to tertiary cardiac care centres and prepare for planned delivery, as well as to establish an appropriate perinatal and postnatal therapeutic plan.},
doi={10.1080/01443615.2021.2000944},
url={https://www.tandfonline.com/doi/abs/10.1080/01443615.2021.2000944},
url={https://doi.org/10.1080/01443615.2021.2000944}

@Article{Gendler2022,
author={Gendler, Y.
and Birk, E.
and Tabak, N.
and of, S. Koton -. International Journal
and 2022, undefined},
title={Vigilance in the Decision-Making Process Regarding Termination of Pregnancy Following Prenatal Diagnosis of Congenital Heart Disease---Application of the },
journal={mdpi.com},
year={2022},
keywords={conflict decision-making model; congenital heart disease; decision-making process; termination of pregnancy; vigilance},
abstract={Publisher's Note: MDPI stays neutral with regard to jurisdictional claims in published maps and institutional affiliations. Abstract: The decision-making process regarding termination of pregnancy following prenatal diagnosis of congenital heart disease is a stressful experience for future parents. Janis and Mann's conflict decision-making model describes seven ideal stages that comprise vigilant information-gathering as an expression of the qualitative decision-making process. In our study, we attempted to determine whether parents who face the decision regarding termination of pregnancy undertake a qualitative decision-making process. Data were collected over 2-year period using structural questionnaires. The sample consisted of two hundred forty participants; sixty-nine (28.75\{\%\}) declared that their decision was to terminate the pregnancy. A significant difference in the quality of the decision-making score was noted between parents who decided to continue with the

pregnancy vs. parents who opted for termination (mean score of 10.15 (5.6) vs. 18.51 (3.9), respectively, $p < 0.001$). Sixty-two (90%) participants within the termination of pregnancy group went through all seven stages of vigilant decision-making process and utilized additional sources for information and consultation. Parents who decided to continue with the pregnancy made swift decisions, often without considering the negative and positive outcomes; this decision-making pattern is considered non-vigilant and ineffective. Identification of future parents at risk of going through an ineffective decision-making process may help health professionals to determine the best way to provide them with information and support.},
doi={10.3390/ijerph19159137},
url={https://www.mdpi.com/1744796},
url={https://doi.org/10.3390/ijerph19159137}

@Article{Kopylov2022,
author={Kopylov, Lital Gordin
and Dekel, Nadav
and Maymon, Ron
and Feldman, Noa
and Zimmerman, Ariel
and Hadas, Dan
and Melcer, Yaakov
and Svirsky, Ran},

title={Prenatally diagnosed isolated perimembranous ventricular septal defect: Genetic and clinical implications},

journal={Wiley Online Library},

year={2022},

month={Apr},

publisher={John Wiley and Sons Ltd},

volume={42},

number={4},

pages={461-468},

abstract={Objective: To evaluate the incidence of chromosomal aberrations and the clinical outcomes following the prenatal diagnosis of isolated perimembranous ventricular septal defect (pVSD). Methods: This retrospective study was composed of a cohort of pregnant women whose fetuses were diagnosed with isolated pVSD. Complete examinations of the fetal heart were performed, as well as a postnatal validation echocardiography follow-up at 1 year of age. The collected data included: spontaneous closure of the pVSD, need for intervention, chromosomal aberrations and postnatal outcome. Results: Fifty-five pregnant women were included in the study. 34/55 (61.8%) of the fetuses underwent prenatal genetic workup which revealed no abnormal results. No dysmorphic features or abnormal neurological findings were detected postnatally in those who declined a prenatal genetic workup during the follow-up period of 2 years. In 25/55 of the cases (45.4%), the ventricular septal defects (VSD) closed spontaneously in utero, whereas in 17 cases of this group (30.9%) the VSD closed during the first year of life. None of the large 3 VSDs cases (>3 mm), closed spontaneously. Conclusion: Prenatally isolated perimembranous VSD has a favorable clinical outcome when classified as small-to-moderate size, children in our cohort born with such findings had no macroscopic chromosomal abnormalities. Key points What's already known about this topic? ♦ The detection rates of perimembranous ventricular septal defect (PVSD) is improving in recent years. What does this study add? ♦ We found no evidence that isolated pVSD is a significant risk factor for chromosomal anomalies. ♦ Isolated small and medium size (<3 mm) PVSD's have a favorable clinical outcome with resolution either prenatally or within the infant's first year of life. This is an open access article under the terms of the Creative Commons Attribution-NonCommercial-NoDerivs License, which

permits use and distribution in any medium, provided the original work is properly cited, the use is non-commercial and no modifications or adaptations are made.},

doi={10.1002/pd.6128},

url={<https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/pd.6128>},

url={<https://doi.org/10.1002/pd.6128>}

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author={Mbbs, Amol A. Moray

and Robertson, Charlene M. T.

and Bond Mn, Gwen Y.

and Abeysekera, Jayani B.

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and Switzer, Heather N.

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and Hornberger, Lisa K.

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title={3rd Trimester Umbilical Arterial Pulsatility Index Is Associated with Neurodevelopmental Outcomes at 2-years in Major Congenital Heart Disease.},

journal={downloads.hindawi.com},

year={2022},

keywords={2 year-Neurodevelopment; Congenital Heart Disease; Middle cerebral artery pulsatility index; Umbilical artery pulsatility index},

abstract={Background},

url={<https://www.researchsquare.com/article/rs-2015109/latest.pdf>}

@Article{Medjedovic2022,

author={Medjedovic, E.

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title={A Single Ventricle Defect: A Case Report from Fetal Echocardiography to Cardiac Surgery},

journal={Article in Donald School Journal of Ultrasound in Obstetrics and Gynecology},

year={2022},

doi={10.5005/jp-journals-10009-1914},

url={https://www.researchgate.net/profile/Edin-Begic/publication/359068062_A_Single_Ventricle_Defect_A_Case_Report_from_Fetal_Echocardiography_to_Cardiac_Surgery/links/6262aae28e6d637bd1f501f7/A-Single-Ventricle-Defect-A-Case-Report-from-Fetal-Echocardiography-to-Cardiac-Surgery.pdf},

url={<https://doi.org/10.5005/jp-journals-10009-1914>}

@Article{Li2022b,

author={Li, T.

and Ma, B.

and Gao, Y.

and Zhang, R.

and Li, P.
and Echocardiography, Z. Da -.
and 2022, undefined},
title={Prenatal diagnosis of total anomalous pulmonary venous connection using 2D and HDlive flow combined with spatiotemporal image correlation},
journal={Wiley Online Library},
year={2022},
publisher={John Wiley and Sons Inc},
keywords={fetus; prenatal diagnosis; spatiotemporal image correlation; total anomalous pulmonary venous connection; two-dimensional; ultrasound},
abstract={Objectives: The objective of this study is to examine the application value of two-dimensional (2D) and high-definition live (HDlive) flow combined with spatiotemporal image correlation (STIC) in diagnosing fetal total anomalous pulmonary venous connection (TAPVC).
Methods: Seventeen cases of fetal TAPVC were diagnosed using 2D and HDlive Flow combined with STIC. These cases were then retrospectively analyzed to examine the value of using 2D and HDlive Flow combined with STIC in the diagnosis of TAPVC.},
doi={10.1111/echo.15429},
url={https://onlinelibrary.wiley.com/doi/abs/10.1111/echo.15429},
url={https://doi.org/10.1111/echo.15429}

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author={Mastromoro, G.
and Hashemian, N. Khaleghi
and Diagnostics, D. Guadagnolo -.
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title={Chromosomal Microarray Analysis in Fetuses Detected with Isolated Cardiovascular Malformation: A Multicenter Study, Systematic Review of the Literature and Meta},
journal={mdpi.com},
year={2022},
month={Jun},
publisher={MDPI},
volume={12},
number={6},
keywords={cardiovascular malformations; chromosomal microarray analysis; fetal malformations; genetic counseling; genetic testing; heart disease; prenatal diagnosis; structural anomalies},
abstract={Citation: Mastromoro, G.; Khaleghi Hashemian, N.; Guadagnolo, D.; Giuffrida, M.G.; Torres, B.; Bernardini, L.; Ventriglia, F.; Piacentini, G.; Pizzuti, A.},
doi={10.3390/diagnostics12061328},
url={https://www.mdpi.com/article/10.3390/diagnostics12061328},
url={https://doi.org/10.3390/diagnostics12061328}}

@Article{Asoglu2022,
author={Asoglu, Mehmet Resit
and Cutting, Elizabeth M.
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and Higgs, Amanda S.
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title={The rate of undetectable genetic causes by Cell-free DNA test in congenital heart defects},
journal={Journal of Maternal-Fetal and Neonatal Medicine},
year={2022},
publisher={Taylor and Francis Ltd.},
volume={35},
number={8},
pages={1484-1490},
keywords={Cell-free DNA; Undetectable genetic causes; congenital heart defect; genetic counseling},
abstract={Objective: The study aimed to estimate the rate of genetic causes that were undetectable by Cell-free DNA (cfDNA) test in prenatally diagnosed congenital heart defect (CHD) cases based on an assumption that cfDNA would accurately detect common aneuploidies including trisomy 21/18/13/45X, and del22q11.2. Methods: This study included prenatally diagnosed CHD cases with diagnostic genetic results. The possibility of false-positive/negative results from cfDNA testing was discarded. Thus, cfDNA results would be positive in common aneuploidies or del22q11.2 and negative in normal diagnostic genetic testing results or other genetic conditions. The rate of genetic causes that were undetectable by cfDNA test was estimated for all cases as well as for CHD subgroups. Results: Of 302 cases, 98 (34.8{\%}) had a type of genetic abnormalities, with 67 having common aneuploidies or del22q11.2 and 31 having other genetic conditions. The rate of genetic causes that were undetectable by cfDNA test in CHD cases was 13.2{\%} among those with assumingly negative cfDNA screen results and 10.3{\%} among the entire study population. These rates were similar between CHD subgroups ($p > .05$). The rate of genetic causes that were undetectable by cfDNA test was higher in the non-isolated cases than in the isolated ones among those with assumingly negative-screen results (20.5{\%} and 9.9{\%}, respectively, $p = .025$). Conclusion: In prenatally diagnosed CDH cases, a significant number of chromosomal abnormalities are still identified after diagnostic testing even if cfDNA screen is negative, and thus it is important to extensively counsel patients with negative cfDNA screen carrying a CHD-affected fetus.},
doi={10.1080/14767058.2020.1757643},
url={https://doi.org/10.1080/14767058.2020.1757643}

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author={Bjoern, Petersen Olav
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and J{o}rgensen, D. S.
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and Sundberg, K.
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title={Prevalence of chromosome disorders in congenital heart defects: a register-based study from Denmark between 2008 and 2018},
journal={Wiley Online Library},
year={2022},
month={Sep},
keywords={chromosomal aberrations; congenital heart defect; genetic disorders; prenatal screening},
doi={10.1002/uog.26075},
url={https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/uog.26075},
url={https://doi.org/10.1002/uog.26075}

@Article{Keleş2022,
author={Keleş, A.
and Yılmaz, O.
and Öyelik -. Gulhane Medical
and 2022, undefined},
title={Characteristics of fetal conotruncal heart anomalies},
journal={cms.gulhanemedj.org},
year={2022},
doi={10.4274/gulhane.galenos.2021.85057},
url={https://cms.gulhanemedj.org/Uploads/Article_52868/GMJ-64-222-En.pdf},
url={https://doi.org/10.4274/gulhane.galenos.2021.85057}}

@Article{Tie2022,
author={Tie, H.
and Ma, B.
and Zhang, D.
and Echocardiography, T. Li -.
and 2022, undefined},
title={Prenatal diagnosis of fetal inferior vena cava malformation using HDlive flow combined with spatiotemporal image correlation},
journal={Wiley Online Library},
year={2022},
month={May},
publisher={John Wiley and Sons Inc},
volume={39},
number={5},
pages={685-690},
keywords={fetus; inferior vena cava malformation; prenatal diagnosis; spatiotemporal image correlation; ultrasound},
abstract={Objectives: This study aimed to examine the application value of high-definition live (HDlive) flow combined with spatiotemporal image correlation (STIC) in the diagnosis of fetal inferior vena cava malformation (IVCM). Methods: Twenty cases of IVCMs were diagnosed using two-dimensional HDlive flow and HDlive flow combined with STIC and retrospectively analyzed to examine the impact of using HDlive flow combined with STIC in the diagnosis of IVCM.},
doi={10.1111/echo.15346},
url={https://onlinelibrary.wiley.com/doi/abs/10.1111/echo.15346},
url={https://doi.org/10.1111/echo.15346}}

@Article{Carberry2022,
author={Carberry, Thomas
and Arzu, Jennifer
and Coons, Dana
and Husain, Nazia
and Gotteiner, Nina
and Webster, Gregory},
title={Postnatal Outcomes in Infants With a History of Fetal Supraventricular Tachycardia},

journal={JACC: Clinical Electrophysiology},
year={2022},
month={Sep},
publisher={Elsevier Inc.},
keywords={atrial flutter; fetal tachycardia; pediatric electrophysiology; supraventricular tachycardia},
abstract={Background: Fetal supraventricular tachycardia (SVT) is rare and proposed predictors of postnatal outcomes in fetal SVT have not been validated. Valid predictors can guide postnatal management. Objectives: The authors correlated fetal characteristics to the incidence of postnatal SVT and compared SVT outcomes in infants with and without a history of fetal SVT. Methods: Mother-fetus dyads with fetal SVT and a structurally normal heart were described and compared with a second cohort of infants with a postnatal diagnosis of SVT. Results: SVT was observed in 78 fetuses and 76 survived to delivery. Maternally administered transplacental antiarrhythmics were used in 49 mother-fetus dyads. Rhythm control was achieved in 37 of 49 (76\%). Among fetuses with intermittent SVT, there was no ventricular dysfunction or hydrops. Postnatal SVT occurred in one-half of infants (37 of 76), and 94\% presented within the first 2 days of life. The following fetal characteristics were associated with postnatal SVT on univariable analysis: sustained SVT (87\% vs 56\%), ventricular dysfunction (41\% vs 15\%), lack of conversion to sinus rhythm (49\% vs 10\%), and earlier gestational age at delivery (37.6 weeks vs 38.9 weeks; $P \leq 0.01$ for each comparison). Compared with infants with a postnatal diagnosis of SVT, infants with a fetal diagnosis presented earlier (median age 0 days vs 17 days; $P < 0.01$) and had a lower incidence ventricular dysfunction at presentation (5\% vs 42\%; $P < 0.01$). Conclusions: One-half of infants with fetal SVT had postnatal SVT, nearly all within 2 days of life. These data and predictors of postnatal SVT may influence parental counseling and postnatal clinical decision-making.},
doi={10.1016/J.JACEP.2022.06.003},
url={https://doi.org/10.1016/J.JACEP.2022.06.003}

@Article{Tsakiridis2022,
author={Tsakiridis, Ioannis G.
and Dagklis, Themistoklis
and Athanasiadis, Apostolos},
title={Obstetrical Monitoring},
journal={Comprehensive Clinical Approach to Diabetes During Pregnancy},
year={2022},
publisher={Springer International Publishing},
pages={133-146},
doi={10.1007/978-3-030-89243-2_8},
url={https://doi.org/10.1007/978-3-030-89243-2_8}}

@Article{Shi2022,
author={Shi, Jia-wei
and Cao, Haiyan
and Hong, Liu
and Ma, Jing
and Cui, Li
and Zhang, Yi
and Song, Xiaoyan
and Liu, Juanjuan
and Yang, Yali

and Lv, Qing
and Zhang, Li
and Wang, Jing
and Xie, Mingxing},
title={Diagnostic yield of whole exome data in fetuses aborted for conotruncal malformations},
journal={Wiley Online Library},
year={2022},
month={Jun},
publisher={John Wiley and Sons Ltd},
volume={42},
number={7},
pages={852-861},
keywords={Key points},
abstract={Objective: We investigated a custom congenital heart disease (CHD) geneset to assess the diagnostic value of whole-exome sequencing (WES) in karyotype-and copy number variation (CNV)-negative aborted fetuses with conotruncal defects (CTDs), and to explore the impact of postnatal phenotyping on genetic diagnosis. Methods: We sequentially analyzed CNV-seq and WES data from 47 CTD fetuses detected by prenatal ultrasonography. Fetuses with either a confirmed aneuploidy or pathogenic CNV were excluded from the WES analyses, which were performed following the American College of Medical Genetics and Genomics recommendations and a custom CHD-geneset. Imaging and autopsy were applied to obtain postnatal phenotypic information about aborted fetuses. Results: CNV-seq identified aneuploidy in 7/47 cases while 13/47 fetuses were CNV-positive. Eighty-five rare deleterious variants in 61 genes (from custom geneset) were identified by WES in the remaining 27 fetuses. Of these, five pathogenic or likely pathogenic variants (PV/LPV) were identified in five fetuses, revealing a 10.6{\%} (5/47) incremental diagnostic yield. Furthermore, RERE: c.2461{_}2472delGGGATGTGGCGA was reclassified as LPV based on postnatal phenotypic data. Conclusion: We have developed and defined a CHD gene panel that can be utilized in a subset of fetuses with CTDs. We demonstrate the utility of incorporating both prenatal and postnatal phenotypic information may facilitate WES diagnostics.},
doi={10.1002/pd.6147},
url={https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/pd.6147},
url={https://doi.org/10.1002/pd.6147}

@Article{Guleroglu2022,
author={Guleroglu, F. Y.
and Ozmen, A. B.
and {\ldots}, M. Ekmez -. Medical Bulletin of
and 2022, undefined},
title={Relationship between Maternal Serum Calcium and Magnesium Levels and Isolated Fetal Echogenic Intracardiac Focus Encountered During Second-trimester},
journal={cms.galenos.com.tr},
year={2022},
volume={60},
pages={40-44},
doi={10.4274/haseki.galenos.2021.7841},
url={https://cms.galenos.com.tr/Uploads/Article_50845/HTB-60-40-En.pdf},
url={https://doi.org/10.4274/haseki.galenos.2021.7841}

@Article{Spurr2022,
author={Spurr, Rebecca R.
and Conwell, Jeffrey A.
and Young, Luciana T.
and Lewin, Mark B.
and Edwards, Lindsay A.
and Arya, Bhawna},
title={Utility of Screening Fetal Echocardiogram Following Normal Anatomy Ultrasound for In Vitro Fertilization Pregnancies},
journal={Pediatric Cardiology},
year={2022},
month={Aug},
publisher={Springer},
volume={43},
number={6},
pages={1349-1353},
keywords={Congenital heart disease; Fetal echocardiogram; Fetal screening; In vitro fertilization},
abstract={In vitro fertilization (IVF) is associated with a higher incidence of congenital heart disease, resulting in universal screening fetal echocardiograms (F-echo) even when cardiac structures on obstetric scan (OB-scan) are normal. Recent studies suggest that when OB-scan is normal, F-echo may add little benefit and increases cost and anxiety. We aim to determine the utility of screening F-echo in IVF pregnancies with normal cardiac anatomy on prior OB-scan. We conducted a retrospective chart review of IVF pregnancies referred for F-echo at the Seattle Children's Hospital between 2014 and 2020. OB-scan results and subspecialty of interpreting physician (Obstetrics = OB; Maternal Fetal Medicine = MFM; Radiology = Rads), F-echoes, and postnatal outcomes were reviewed. Cardiac anatomy on OB-scans was classified as complete if 4-chamber and outflow-tract views were obtained. Supplemental views (three-vessel and sagittal aortic arch views) on OB-scan were also documented. Of 525 IVF referrals, OB-scan reports were available for review in 411. Normal anatomy was demonstrated in 304 (74%) interpreted by OB (128; 42%), MFM (80; 26%), and Rads (96; 32%). F-echo was normal in 278 (91%). Of the 26 abnormal F-echo, none required intervention (17 muscular and 5 perimembranous ventricular septal defects, and 4 minor valve abnormalities). There was no difference in OB-scan accuracy for identifying normal cardiac anatomy when comparing 4-chamber and outflow-tract views vs. addition of supplemental views (91% vs 92% normal F-echo; $p > 0.1$). Evaluation of OB-scan accuracy by interpreting physician subspecialty demonstrated normal F-echo in 95%, 85%, and 92% ($p = 0.95$) as read by OB, MFM, and Rads, respectively. A majority of IVF referrals with normal cardiac anatomy visualized on OB-scan using 4-chamber and outflow-tract views resulted in normal F-echo, regardless of interpreting physician subspecialty or addition of supplemental views. Of the minority with abnormal F-echo, none required intervention. Consideration should be given to the cost/benefit of screening F-echo for the indication of IVF if normal cardiac anatomy is demonstrated on OB-scan.},
doi={10.1007/S00246-022-02857-5},
url={https://doi.org/10.1007/S00246-022-02857-5}

@Article{Poenaru2022,
author={Poenaru, Mircea-Octavian
and Hamoud, Bashar Haj
and Sima, Romina-Marina
and Valcea, Ionut-Didel
and Chicea, Radu
and Ples, Liana},

title={Persistent Left Superior Vena Cava Significance in Prenatal Diagnosis---Case Series},
journal={mdpi.com},
year={2022},
month={Jul},
publisher={MDPI},
volume={2022},
number={14},
pages={4020-4020},
keywords={congenital heart disease; congenital venous anomaly; persistent left superior vena cava; prenatal diagnosis},
abstract={Citation: Poenaru, M.-O.; Hamoud, B.H.; Sima, R.-M.; Valcea, I.-D.; Chicea, R.; Ples, L. Persistent Left Superior Vena Cava Significance in Prenatal Diagnosis-Case Series. Abstract: The persistent left superior vena cava (PLSVC) is a congenital heart anomaly reported in 0.3-0.5{\%} of the general population and can be associated with congenital heart diseases in up to 8{\%} of cases. Prenatal identification of PLSVC is important to prompt an extended cardiac and extracardiac fetal examination. We retrospectively reevaluated anomaly scans performed in our unit in a 2-year interval according to the national guidelines to evaluate the incidence of PLSVC and its association with prenatal morbidity. In our population, the incidence of PLSVC was 0.31{\%}, and we found a low association with cardiac and extracardiac anomalies. The standard sections (three-vessel and trachea view, four-chamber view and outflow tract's view) are insufficient to exclude cardiac anomalies whenever PLSVC is found. In our case series, only one newborn required postnatal surgery for total pulmonary vein anomaly, and at 2 years of life all babies had a normal evolution. Prenatal diagnosis of PLSVC can raise counseling issues; therefore, awareness of its good outcome when isolated and need for an extended examination to rule out other anomalies is very important.},
doi={10.3390/jcm11144020},
url={https://www.mdpi.com/article/10.3390/jcm11144020},
url={https://doi.org/10.3390/jcm11144020}

@Article{Hornberger2021,
author={Hornberger, Lisa K.
and McBrien, Angela},
title={Extracardiac Pathology in Fetal Heart Disease: What You See Is Not Always What You Get},
journal={Journal of the American College of Cardiology},
year={2021},
month={Dec},
publisher={Elsevier Inc.},
volume={78},
number={23},
pages={2323-2325},
keywords={congenital heart disease; extracardiac anomalies; fetal cardiac magnetic resonance; fetal heart; structural brain anomalies; ultrasound},
doi={10.1016/J.JACC.2021.09.1359},
url={https://doi.org/10.1016/J.JACC.2021.09.1359}

@Article{Holmes2021,
author={Holmes, Kathryn W.
and Huang, Jennifer H.
and Gutshall, Kristine

and Kim, Amanda
and Ronai, Christina
and Madriago, Erin J.},
title={Fetal counseling for congenital heart disease: is communication effective?},
journal={Journal of Maternal-Fetal and Neonatal Medicine},
year={2021},
publisher={Taylor and Francis Ltd.},
keywords={Congenital heart disease; fetal counseling},
abstract={Purpose: To assess the accuracy of maternal understanding of fetal cardiac defects following initial fetal counseling. Methods: Pregnant women with a fetal diagnosis of congenital heart disease (CHD) were surveyed regarding understanding of their fetus's heart defect. The survey asked: (1) for a description of the heart condition; (2) how confident they were in the diagnosis; (3) whether their fetus would require heart surgery. Two fetal cardiologists evaluated the maternal qualitative description. Partners were excluded from the study. Results: Fifty-one participants consented and 39 completed the survey. Mean age was 31 years, 60{\%} had some college level or post-graduate education, 48{\%} had Medicaid insurance, and 81{\%} were Caucasian. More than three-quarters of participants, stated they had either "quite a bit" or "very much" understanding of their fetus's diagnosis. Maternal assessment matched the physician's assessment of accuracy with 77{\%} (N = 30) demonstrating either "quite a bit" or a "very accurate" description of the diagnosis. All women correctly understood if their fetus would require heart surgery. Highest level of maternal education positively correlated with the accuracy of diagnosis (regression coefficient 0.48, $p < .002$). However, confidence in the diagnosis was independent of both education (0.30, $p = .167$) and maternal age (-0.03 , $p = .234$). Conclusions: Fetal counseling is effective in conveying anatomy and the need for surgery; however, accuracy amongst women with lower levels of education and maternal confidence in understanding can be improved.},
doi={10.1080/14767058.2021.1874909},
url={https://doi.org/10.1080/14767058.2021.1874909}

@Article{Veronese2021,
author={Veronese, Paola
and Bertelli, Francesco
and Cattapan, Claudia
and Andolfatto, Matteo
and Gervasi, Maria Teresa
and Vida, Vladimiro L.},
title={Three-Dimensional Printing of Fetal Heart With d -Transposition of the Great Arteries From Ultrasound Imaging Data},
journal={World Journal for Pediatric and Congenital Heart Surgery},
year={2021},
month={Mar},
publisher={SAGE Publications},
volume={12},
number={2},
pages={291-292},
abstract={We reconstructed and printed a 3D model of the fetal heart affected by d-transposition of the great arteries from prenatal ultrasound images. Our 3D model revealed to be very helpful in showing the basic anatomical features of fetal complex Congenital Heart Disease (CHD) and represents an interesting additional diagnostic tool to the current standard imaging armamentarium, improving the quality of prenatal parental counseling.},
doi={10.1177/2150135120947687},

url={<https://doi.org/10.1177/2150135120947687>}

@Article{Nagata2021,
author={Nagata, Hazumu
and Yamamura, Kenichiro
and Matsuoka, Ryohei
and Kato, Kiyoko
and Ohga, Shouichi},
title={Diagnosis of congenital heart disease by early and second-trimester fetal echocardiography},
journal={Wiley Online Library},
year={2021},
month={Jan},
publisher={NLM (Medline)},
volume={64},
number={1},
pages={e15098-e15098},
keywords={fetal congenital heart disease; maternal congenital heart disease; transitional care},
abstract={The number of women with congenital heart disease (CHD) reaching reproductive age has been increasing. Many women with CHDs are desirous of pregnancy, but they face issues regarding preconception, antepartum, and post-partum management. On the other hand, the fetal diagnosis of CHD has improved with advances in the technique and equipment for fetal echocardiography. Recently, experiences with fetal intervention have been reported in patients with severe CHD, such as critical aortic stenosis. Nevertheless, some types of CHD are challenge to diagnose prenatally, resulting in adverse outcomes. Medical care is part of the transitional care for women and fetuses with CHD during the perinatal period. Pre-conceptual and prenatal counseling play an important role in transitional care. Sex and reproductive education need to be performed as early as possible. We herein review the current status, important issues to be resolved, and the future of maternal and fetal CHD to relevant caregivers.},
doi={10.1111/ped.15098},
url={<https://onlinelibrary.wiley.com/doi/abs/10.7863/jum.2012.31.4.563>},
url={<https://doi.org/10.1111/ped.15098>}

@Article{Digital2021,
author={Digital, Jefferson
and Commons Phase, Jefferson Digital
and Houlihan, Taylor Hartzel
and Rychik, Jack},
title={Factors influencing efficacy and satisfaction in prenatal counseling for families with single ventricle heart disease},
year={2021},
url={https://jdc.jefferson.edu/si_ctr_2023_phase1/10/}

@Article{Day2021,
author={Day, Thomas G.
and Kainz, Bernhard
and Hajnal, Jo

and Razavi, Reza
and Simpson, John M.
and Children, London
and Thomas, St},
title={Artificial intelligence, fetal echocardiography, and congenital heart disease},
journal={Wiley Online Library},
year={2021},
month={May},
publisher={John Wiley and Sons Ltd},
volume={41},
number={6},
pages={733-742},
keywords={41; 733-742 wileyonlinelibrary.com/journal/pd-733; Prenatal Diagnosis 2021},
abstract={There has been a recent explosion in the use of artificial intelligence (AI), which is now part of our everyday lives. Uptake in medicine has been more limited, although in several fields there have been encouraging results showing excellent performance when AI is used to assist in a well-defined medical task. Most of this work has been performed using retrospective data, and there have been few clinical trials published using prospective data. This review focuses on the potential uses of AI in the field of fetal cardiology. Ultrasound of the fetal heart is highly specific and sensitive in experienced hands, but despite this there is significant room for improvement in the rates of prenatal diagnosis of congenital heart disease in most countries. AI may be one way of improving this. Other potential applications in fetal cardiology include the provision of more accurate prognoses for individuals, and automatic quantification of various metrics including cardiac function. However, there are also ethical and governance concerns. These will need to be overcome before AI can be widely accepted in mainstream use. It is likely that a familiarity of the uses, and pitfalls, of AI will soon be mandatory for many healthcare professionals working in fetal cardiology. Key Points What's already known about this topic? ♦ Artificial intelligence (AI) is rapidly becoming part of everyday life, and interest is growing in its use in improving medical outcomes. ♦ AI may be a useful tool in fetal cardiology to improve rates of antenatal diagnosis of congenital heart disease, as well as other potential benefits. ♦ Potential pitfalls exist however, and future clinicians will likely need to have a thorough understanding of the risks and benefits of AI. What does this study add? ♦ This review article summarizes the fundamentals of AI, the potential uses of AI in fetal cardiology, and what the future may hold. This is an open access article under the terms of the Creative Commons Attribution License, which permits use, distribution and reproduction in any medium, provided the original work is properly cited.},
doi={10.1002/pd.5892},
url={https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/pd.5892},
url={https://doi.org/10.1002/pd.5892}

@Article{Cohen2021,
author={Cohen, Jennifer
and Arya, Bhawna
and Donofrio, Mary T.
and Harrington, Jamie K.
and Ho, Deborah Y.
and Hogan, Whitnee J.
and Hornberger, Lisa K.
and Killen, Stacy A.
and Michelfelder, Erik
and Moon-Grady, Anita J.
and Patel, Sheetal R.

and Quezada, Emilio
and ronai, christina
and Sanchez Mejia, Aura A.
and Schidlow, David N.

and Srivastava, Shubhika},

title={Abstract 11120: Congenitally Corrected Transposition of the Great Arteries - Fetal Diagnosis, Associations and Natural History: A Fetal Heart Society Research Collaborative Study},

journal={Circulation},

year={2021},

month={Nov},

publisher={Ovid Technologies (Wolters Kluwer Health)},

volume={144},

number={Suppl{1}},

abstract={Introduction: Congenitally corrected transposition of the great arteries (ccTGA) is rare with varied associated cardiac defects and rhythm abnormalities. We aimed to describe the natural history, associated anomalies and prenatal outcome in a cohort of prenatally diagnosed patients in which biventricular repair is anticipated. Methods: A retrospective cohort study was conducted via the Fetal Heart Society Research Collaborative. All fetuses with ccTGA encountered at 15 North American cardiac centers between 1/2004-7/2020 were identified. Fetuses with a hypoplastic ventricle precluding biventricular repair were excluded. Data is presented as median (interquartile range). Results: Inclusion criteria were met in 139 fetuses who were diagnosed with ccTGA at 24 (21-29) weeks. There was a family history of congenital heart disease in 12{%. Maternal diabetes was present in 10{%. Prenatal genetic testing in 50 pregnancies was normal. Excluding 14 fetuses with heterotaxy, extracardiac anomalies were observed in 9/125 (7.1{%). Associated cardiac/extracardiac defects are detailed in table 1. Fetal atrioventricular block (AVB) was present in 17 fetuses (12{%), diagnosed at a median of 26 (23-28), range 20-36 weeks. Two fetuses had SVT; 1 self-resolved and 1 during labor. Change occurred during follow-up in 34 fetuses, most commonly in severity of tricuspid regurgitation (6 improved, 4 worsened) and worsening of pulmonary stenosis (n=5). There were 112 live births, 17 terminations, 2 fetal deaths (FD) and 8 lost to follow-up. Both fetuses with FD had AVB, making the intrauterine mortality associated with fetal AVB 11.8{%} (2/17). Conclusions: This study represents the largest cohort to date of fetuses with ccTGA. Fetal ccTGA is associated with a spectrum of cardiac defects that may evolve, with extracardiac pathology occurring in 7.1{%. AVB is found in 12{%} of fetuses and is a risk factor for FD. This data informs fetal counseling and can aid in delivery planning and perinatal care.},

doi={10.1161/CIRC.144.SUPPL_1.11120},

url={https://doi.org/10.1161/CIRC.144.SUPPL_1.11120}

@Article{HernándezHern2021,

author={Hern{a}ndezHern, D.

and P{e}rez, Hern{a}ndez

and Garcia Delgado, R.

and Garcia Rodriguez, R.

and Amaro Acosta, A.

and Ortega, I. C.

and Segura Gonzalez, J.

and Benitez Delgado, T.

and Medina Castellano, M.},

title={VP21.13: Late diagnosis of a complex heart disease due to COVID-19 pandemic: the key role of prenatal monitoring and ultrasound anomaly assessment},

journal={Ultrasound in Obstetrics {&} Gynecology},

year={2021},
month={Oct},
publisher={Wiley},
volume={58},
number={S1},
pages={186-187},
abstract={Objectives: To analyse the cases of fetuses with persistent left superior vena cava (PLSVC) at our centre, evaluating prenatal, perinatal and postnatal data. Methods: This is retrospective observational study carried at a tertiary hospital which included all cases of with PLSVC diagnosed or referred to our Prenatal Diagnosis Unit from 2015 to 2021. Results: A total of 46 fetuses diagnosed with PLSVC were found. However, due to loss of follow-up, we performed the analysis at 37 patients. The diagnosis of PLSVC was established in 11{\%} of patients at < 18 weeks, 81{\%} at 18-23 weeks and in 8{\%} at > 23 weeks. Isolated persistent left superior vena cava was in 32{\%} and in 68{\%} of the patients presented PLSVC together with other ultrasound findings, being the most frequent pathologies coarctation of the aorta, pathologies of cardiac axis and heterotaxy syndrome or central nervous system abnormalities. In the 13{\%} we found the absence of the right superior vena cava. Invasive testing was performed on 12 patients, all of which 50{\%} were abnormal. We did not do invasive test in isolated PLSVC. 19{\%} of the patients decided to carry out termination of pregnancy.},
doi={10.1002/UOG.24343},
url={https://doi.org/10.1002/UOG.24343}

@Article{Afshar2021,
author={Afshar, Yalda
and Whitnee, ;.
and Hogan, J.
and Conturie, Charlotte
and Sunderji, Sherzana
and Duffy, Jennifer Y.
and Peyvandi, Shabnam
and Boe, Nina M.
and Melber, Dora
and Fajardo, Viviana M.
and Tandel, Megha D.
and Holliman, Kerry
and Kwan, Lorna
and Satou, Gary
and Moon-Grady, Anita J.},
title={Multi-Institutional Practice-Patterns in Fetal Congenital Heart Disease Following Implementation of a Standardized Clinical Assessment and Management Plan},
journal={Am Heart Assoc},
year={2021},
publisher={American Heart Association Inc.},
volume={10},
number={15},
pages={21598-21598},
keywords={Cesarean; Fetal CHD; Obstetrics; Prenatal congenital heart disease; SCAMP},
abstract={BACKGROUND: Prenatal diagnosis of congenital heart disease has been associated with early-term delivery and cesarean delivery (CD). We implemented a multi-institutional standardized clinical assessment and management plan (SCAMP) through the University of California Fetal-Maternal Consortium. Our objective was to decrease early-term (37-39 weeks) delivery and CD

in pregnancies complicated by fetal congenital heart disease using a SCAMP methodology to improve practice in a high-risk and clinically complex setting.},
doi={10.1161/JAHA.121.021598},
url={https://www.ahajournals.org/doi/abs/10.1161/JAHA.121.021598},
url={https://doi.org/10.1161/JAHA.121.021598}

@Article{Asrani2021,
author={Asrani, Priyanka
and Friedman, Kevin},
title={Prenatal Detection of Congenital Heart Disease: the Past, Present, and Future},
journal={Current Treatment Options in Cardiovascular Medicine},
year={2021},
month={Feb},
publisher={Springer},
volume={23},
number={2},
keywords={Congenital heart disease; Fetal cardiology; Prenatal diagnosis},
abstract={Purpose of review: Prenatal diagnosis of congenital heart disease (CHD) is continuously evolving with each passing decade. Early efforts in fetal cardiology focused on identifying CHD in mid-gestation and understanding of fetal circulation in pathologic conditions. Improving prenatal detection rates for CHD remains an ongoing challenge and increasingly the field of fetal cardiology is moving to not only diagnosing CHD prenatally but also assessing the impact of prenatal diagnosis of CHD outcomes. Future directions include earlier diagnosis of fetal CHD, improved diagnostic rates, widespread sonographer education, and a better understanding of antenatal factors that impact outcomes. Our goal in this review is to describe the past, present, and future of prenatal diagnosis of CHD. Recent findings: There has been a steady improvement in the prenatal diagnosis rate for CHD; however, there remains a significant variation between countries and within the USA. Prenatal diagnosis of CHD allows for counseling and delivery planning in those fetuses with critical CHD, thereby providing parents with resources and important tools while dealing with a challenging situation of carrying a child with heart disease and helping them with important decisions for their family and their future. There are several specific conditions which continue to pose a challenge from a diagnostic standpoint as they may appear mild at the time of initial diagnosis and may be missed but progress through the pregnancy and lead to significant CHD in the neonatal period. Summary: In summary, continued efforts aimed at collaborative research and education are needed in order to be able to improve CHD detection rates. We need to cautiously assess lesions that appear minor in mid-gestation but have the potential to progress in late gestation. Earlier detection of CHD by means of a transvaginal or a first-trimester fetal echocardiogram may further help families with delivery planning and decision-making.},
doi={10.1007/S11936-020-00886-Y},
url={https://doi.org/10.1007/S11936-020-00886-Y}

@Article{Jansen2021,
author={Jansen, F.
and Velzen, C. van
and {Idots}, E. Pajkrt -. {Idots} in Obstetrics {&}
and 2014, undefined},
title={OP 01.01: Head circumference growth in isolated fetal heart defects},
journal={Wiley Online Library},

year={2021},
month={Oct},
publisher={Wiley},
volume={58},
number={S1},
pages={188-188},

abstract={Virtual poster abstracts of this review was to summarise available evidences concerning prenatal diagnosis and neonatal outcomes of LBCV anomalies (LBCVA) with particular interest to the risk of congenital heart disease (CHD), extracardiac (ECA) or genetic abnormalities. Methods: The web-based databases were extensively searched and 13 studies were retrieved from literature, with a total of 308 cases of LBCVA. Six cases from our series of pregnancies were added to cases from literature. Pooled proportions were calculated for risk of associated CHD, ECA or genetic abnormalities and obstetric outcomes. Results: 308 cases of LBCVA were collected, of which: 107 abnormal courses; 177 absent LBCV with a left superior vena cava (5 with absent right SVC); 20 LBCV dilation; 2 double LBVC. Of 308 cases of LBCVA 234 (75,9\%) had an associated CHD (70,6\% in retroesophageal, 65,5\% in subaortic and 4,3\% in intrathymic LBCV). Absent and double LBCV were always associated with CHD. ECAs were reported in 10 cases (3.2\%). Genetic data were available in 19 fetuses; in 6 there was a genetic anomaly, with 3 cases of RASopathy. Neonatal outcomes were available for 64 fetuses of which 51 (79,7\%) were normal. Conclusions: LBCVA are generally isolated defects with uncertain prevalence and rare prenatal detection. Unlike intrathymic forms, absent, double, subaortic, retroesophageal and dilated LBCV are frequently associated with CHD. Data on genetic abnormalities are scarce with higher risk for retroesophageal and subaortic courses. Neonatal outcomes were available for a minority of cases and it was favourable in most fetuses with intrathymic course, absent or dilated LBCV. VP22.03 Abstract withdrawn VP22.04 Prenatal diagnosis, genetic analysis and prognosis of isolation of aortic brachiocephalic artery},
doi={10.1002/uog.24349},
url={https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/uog.13637},
url={https://doi.org/10.1002/uog.24349}

@Article{Gao2021,
author={Gao, Shuang
and Han, Jiancheng
and Yu, Shaomei
and Guo, Yong
and Ruan, Yanping
and Fu, Yuwei
and Hao, Xiaoyan
and Wang, Xin
and Wang, Siyu
and Zhou, Xiaoxue
and Shang, Jianfeng
and Zhang, Ye
and Li, Tianjing
and Hao, Xiuxiu
and He, Yihua},

title={Comparison of fetal echocardiogram with fetal cardiac autopsy findings in fetuses with congenital heart disease},
journal={Journal of Maternal-Fetal and Neonatal Medicine},
year={2021},

publisher={Taylor and Francis Ltd.},
volume={34},
number={23},
pages={3844-3850},
keywords={Cardiac autopsy; comparison; congenital heart disease; fetal echocardiography},
abstract={Objective: Although studies have compared fetal echo results with autopsy findings, investigations that compared multiple categories of congenital heart disease (CHD) are lacking. This study, therefore, aimed to compare fetal echocardiographic diagnoses with cardiac autopsy findings and evaluate the diagnostic accuracy of fetal echocardiography (FE). Methods: One hundred seventy-one specimens from fetuses diagnosed with CHD were collected after termination of pregnancy, and fetal autopsies were performed. FE and autopsy diagnoses were compared and the degree of their correspondence was categorized as "complete agreement" (FE results were in accordance with autopsy findings), "minor discrepancies" (autopsies verified the main FE diagnoses but added and/or revised some minor information), or "discordance" (autopsy findings were different from the primary diagnoses of FE). Results: The "complete agreement" group accounted for 87.1% (149/171) of the total specimens. In 11.7% (20/171) of cases, autopsies disclosed new deformities and/or revised some echo results (minor discrepancies group). Minor abnormalities were frequently embodied in small septal defects and vascular malformations. A rare malformation of common pulmonary vein atresia was confirmed by autopsy in two fetuses, but both were misdiagnosed by FE (discordance group). Conclusions: Fetal echocardiographic diagnoses were mostly consistent with autopsy findings. The diagnostic discrepancies mainly consisted of rare cases and minor abnormalities missed or misdiagnosed by FE. Autopsies may help confirm, modify, or add information to prenatal echo results. They may also help sonographers have a better understanding of the anatomic structures of CHD, especially for rare lesions, which could further improve the diagnostic accuracy and integrity of FE.},
doi={10.1080/14767058.2019.1700498},
url={https://doi.org/10.1080/14767058.2019.1700498}

@Article{Lee2021,
author={Lee, Caroline K.
and Michelfelder, Erik C.
and Singh, Gautam K.},
title={Fetal echocardiography},
journal={Clinical Maternal-Fetal Medicine Online},
year={2021},
month={Sep},
publisher={CRC Press},
address={London},
pages={64.1-64.19},
doi={10.1201/9781003222590-57},
url={https://www.taylorfrancis.com/books/9781003222590/chapters/10.1201/9781003222590-57},
url={https://doi.org/10.1201/9781003222590-57}

@Article{Freud2021,
author={Freud, Lindsay R.
and Hornberger, Lisa K.},
title={Fetal Echocardiography},
journal={Echocardiography in Pediatric and Congenital Heart Disease},

year={2021},
month={Dec},
publisher={Wiley},
pages={924-963},
doi={10.1002/9781119612858.CH44},
url={https://onlinelibrary.wiley.com/doi/10.1002/9781119612858.ch44},
url={https://doi.org/10.1002/9781119612858.CH44}

@Article{Sun2021,
author={Sun, Liqun
and Lee, Fu-Tsuen
and van Amerom, Joshua F. P.
and Freud, Lindsay
and Jaeggi, Edgar
and Macgowan, Christopher K.
and Seed, Mike},
title={Update on fetal cardiovascular magnetic resonance and utility in congenital heart disease},
journal={Journal of Congenital Cardiology},
year={2021},
month={Dec},
publisher={Springer Science and Business Media LLC},
volume={5},
number={1},

abstract={There are 0.6-1.9{\%} of US children who were born with congenital heart malformations. Clinical and animal studies suggest that abnormal blood flow forces might play a role in causing these malformation, highlighting the importance of understanding the fetal cardiovascular fluid mechanics. We performed computational fluid dynamics simulations of the right ventricles, based on four-dimensional ultrasound scans of three 20-wk-old normal human fetuses, to characterize their flow and energy dynamics. Peak intraventricular pressure gradients were found to be 0.2-0.9 mmHg during systole, and 0.1-0.2 mmHg during diastole. Diastolic wall shear stresses were found to be around 1 Pa, which could elevate to 2-4 Pa during systole in the outflow tract. Fetal right ventricles have complex flow patterns featuring two interacting diastolic vortex rings, formed during diastolic E wave and A wave. These rings persisted through the end of systole and elevated wall shear stresses in their proximity. They were observed to conserve approximately 25.0{\%} of peak diastolic kinetic energy to be carried over into the subsequent systole. However, this carried-over kinetic energy did not significantly alter the work done by the heart for ejection. Thus, while diastolic vortexes played a significant role in determining spatial patterns and magnitudes of diastolic wall shear stresses, they did not have significant influence on systolic ejection. Our results can serve as a baseline for future comparison with diseased hearts.},
doi={10.1186/S40949-021-00059-X},
url={https://doi.org/10.1186/S40949-021-00059-X}

@Article{Mullen2021,
author={Mullen, McKay
and Zhang, Angela
and Lui, George K.
and Romfh, Anitra W.
and Rhee, June Wha

and Wu, Joseph C.},
title={Race and Genetics in Congenital Heart Disease: Application of iPSCs, Omics, and Machine Learning Technologies},
journal={Frontiers in Cardiovascular Medicine},
year={2021},
month={Feb},
publisher={Frontiers Media S.A.},
volume={8},
keywords={congenital heart disease; disease modeling; disparity; genomics; iPSC; race},
abstract={Congenital heart disease (CHD) is a multifaceted cardiovascular anomaly that occurs when there are structural abnormalities in the heart before birth. Although various risk factors are known to influence the development of this disease, a full comprehension of the etiology and treatment for different patient populations remains elusive. For instance, racial minorities are disproportionately affected by this disease and typically have worse prognosis, possibly due to environmental and genetic disparities. Although research into CHD has highlighted a wide range of causal factors, the reasons for these differences seen in different patient populations are not fully known. Cardiovascular disease modeling using induced pluripotent stem cells (iPSCs) is a novel approach for investigating possible genetic variants in CHD that may be race specific, making it a valuable tool to help solve the mystery of higher incidence and mortality rates among minorities. Herein, we first review the prevalence, risk factors, and genetics of CHD and then discuss the use of iPSCs, omics, and machine learning technologies to investigate the etiology of CHD and its connection to racial disparities. We also explore the translational potential of iPSC-based disease modeling combined with genome editing and high throughput drug screening platforms.},
doi={10.3389/FCVM.2021.635280/FULL},
url={https://doi.org/10.3389/FCVM.2021.635280/FULL}

@Article{Tidrenczel2021,
author={Tidrenczel, Zsolt
and Hajdu, Julia
and {Simonyi}
and {At{\e}n{\e}}
and Szab{o}, Istv{a}n
and {Acs}
and {N{a}ndor}
and Demeter, J{a}nos
and {Beke}
and {Art{u}r}},

title={Trends in the prenatal diagnosis of trisomy 21 show younger maternal age and shift in the distribution of congenital heart disease over a 20-year period},
journal={Wiley Online Library},
year={2021},
month={Jun},
publisher={John Wiley and Sons Inc},
volume={185},
number={6},
pages={1732-1742},
keywords={Down syndrome; congenital heart disease; fetopathology; prenatal ultrasound; septal defects},

abstract={Prenatal testing has changed greatly over the past two decades, which may affect the diagnosis of congenital heart disease (CHD) in Down syndrome. The present study aimed to analyze

changes in the prevalence and distribution of CHD diagnosed via ultrasonography and fetopathology in 462 fetuses with trisomy 21 between two consecutive 10-year periods (1999-2018), as well as the associations between CHDs, ultrasound markers, and extracardiac malformations. Overall, the frequency of cardiovascular malformations in trisomy 21 was 27.7 and 26.5{\\%}, and ultrasound identified 70 and 62{\\%} of CHDs during these periods. A profound increase in first-trimester ultrasound findings and associated anomalies with CHDs (ventricular septal defect, Tetralogy of Fallot) since 2009 were observed. Second-trimester non-structural heart abnormalities were associated with ultrasound anomalies (74{\\%}) and major extracardiac malformations (42.9{\\%}). During both study periods, mothers carrying fetuses with CHD were significantly younger than those without CHD ($p = 0.038$, $p = 0.009$, respectively). Comparing the two 10-year periods, there were no changes in the prevalence and detection of CHDs. Trend analysis revealed that, although the frequency of CHD remained stable, the diagnostic spectrum had shifted between the study periods. Detection of nonstructural heart abnormalities necessitates detailed follow-up for cardiac/extracardiac malformations and chromosomal disorders. K E Y W O R D S congenital heart disease, Down syndrome, fetopathology, prenatal ultrasound, septal defects},
doi={10.1002/ajmg.a.62162},
url={https://onlinelibrary.wiley.com/doi/abs/10.1002/ajmg.a.62162},
url={https://doi.org/10.1002/ajmg.a.62162}

@Article{Cao2021,
author={Cao, Li
and Du, Yan
and Zhang, Mo
and Wang, Feng
and Zhao, Jian-Yuan
and Ren, Yun-Yun
and Gui, Yong-Hao},
title={High maternal blood lipid levels during early pregnancy are associated with increased risk of congenital heart disease in offspring},
journal={Wiley Online Library},
year={2021},
month={Oct},
publisher={John Wiley and Sons Inc},
volume={100},
number={10},
pages={1806-1813},
keywords={Apo-B, apolipoprotein-B; BMI, body mass index; CHD, congenital heart disease; HDL-C, high-density lipoprotein cholesterol; HbA1c, hemoglobin A1c; Hcy, homocysteine; LDL-C, low-density lipoprotein cholesterol; TC, total cholesterol; TG, triglyceride; contributed equally to this work
Abbreviations: Apo-A1, apolipoprotein-A1},
abstract={This is an open access article under the terms of the Creative Commons Attribution-NonCommercial-NoDerivs License, which permits use and distribution in any medium, provided the original work is properly cited, the use is non-commercial and no modifications or adaptations are made. Abstract Introduction: This study aimed to investigate whether maternal blood lipid levels dur},
doi={10.1111/aogs.14225},
url={https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1111/aogs.14225},
url={https://doi.org/10.1111/aogs.14225}

@Article{Kaymak2021,
author={Kaymak, Didem
and Alpay, Verda
and Ba{\c{s}}{\i}b{\u}y{\u}k, Zafer
and Davuto{\u{g}}lu, Ebru Al{\i}c{\i}
and Madaz{\i}, Riza},
title={Prenatal Diagnosis of 8p23 Deletion Syndrome by Single Nucleotide Polymorphism Microarray},
journal={Journal of Fetal Medicine},
year={2021},
month={Dec},
publisher={Springer Science and Business Media LLC},
volume={8},
number={4},
pages={315-319},
doi={10.1007/S40556-021-00322-6},
url={https://doi.org/10.1007/S40556-021-00322-6}}

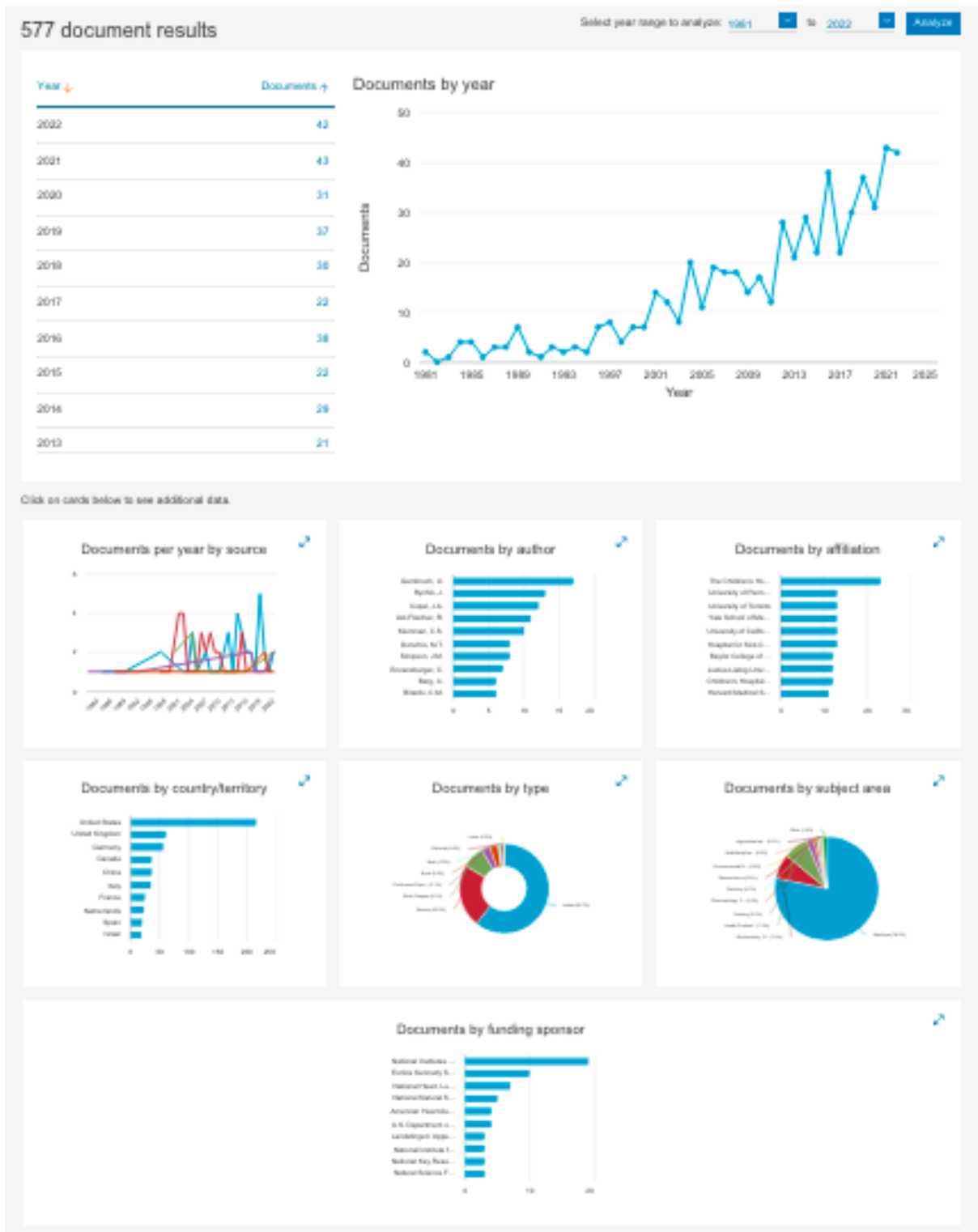
@Article{Jin2021,
author={Jin, Xiao
and Ni, Wei
and Wang, Guolan
and Wu, Qin
and Zhang, Jun
and Li, Guoju
and Jiao, Na
and Chen, Wenjing
and Liu, Qing
and Gao, Li
and Xing, Quansheng},
title={Incidence and risk factors of congenital heart disease in Qingdao: a prospective cohort study},
journal={BMC Public Health},
year={2021},
month={Dec},
publisher={BioMed Central Ltd},
volume={21},
number={1},
keywords={Congenital heart defects; Epidemiologic studies; Incidence; Regression analysis; Risk factors},
abstract={Background: Many studies have been conducted to assess the incidence of congenital heart disease (CHD). However, results were greatly inconsistent among these studies with a broad range of findings. Methods: A prospective census-based cohort study was conducted in Qingdao, China, from August 1, 2018 to April 30, 2019. All of the local registered pregnant women were continuously investigated and followed from 15 to 20 weeks of gestation to delivery, tracking the CHD cases in both the fetal and neonatal stages. A logistic regression model was applied to assess the association between CHD and possible risk factors. Results: The positive rate of prenatal CHD screening was 14.36 per 1000 fetuses and the incidence of CHD was 9.38 per 1000 live births. Results from logistic regression indicated that, living in the countryside (odds ratio, (OR): 0.771; 95{\%} confidence interval, (CI): 0.628--0.946) and having a childbearing history (OR: 0.802; 95{\%}CI: 0.676--0.951) were negatively associated with CHD. However, twin pregnancy (OR: 1.957, 95{\%} CI: 1.245--3.076), illness in the first trimester (OR: 1.306; 95{\%} CI: 1.048--1.628), a family history of CHD (OR: 7.156;

95% CI: 3.293--15.552), and having a child with a birth defect (OR: 2.086; 95% CI: 1.167--3.731) were positively associated with CHD. Conclusion: CHD is a serious health problem in Qingdao. The CHD incidence found in this study was similar to existing research. The positive rate of prenatal CHD screening was higher than the incidence of neonatal CHD. Moreover, CHD risk factors were identified in our study, and our findings may have great implications for formation CHD intervention strategies.},
doi={10.1186/S12889-021-11034-X},
url={https://doi.org/10.1186/S12889-021-11034-X}

@Article{Evans2021,
author={Evans, William N.
and Acherman, Ruben J.
and Restrepo, Humberto},
title={Prenatal dia},
year={2021},
publisher={Taylor and Francis Ltd.},
keywords={Congenital heart disease; fetal comorbidities; fetal echocardiography; prenatal detection;
syndromes; termination of pregnancy},
abstract={Objective: We investigated the relationship between prenatal detection of significant congenital heart disease and elective termination of pregnancy over time in Nevada. Methods: We identified those prenatally or post-natally diagnosed with significant congenital cardiovascular malformations in Nevada with birth dates or estimated delivery dates between July 2012 and June 2021. Results: We identified 1246. Of 1246, 69 underwent fetal demise, 42 had elective termination, and 1135 were live-born. Of the 1135 live-born, 1090 had prenatal care, of which 718 (66%) overall had a prenatal diagnosis of significant congenital heart disease. However, prenatal detection statistically significantly increased over time from 45 to 82%, $p = .00001$. Termination of pregnancy averaged 10% of those identified within the legal timeframe, and the rate did not statistically significantly increase with increasing prenatal detection rates, $p = .56$. Of the 42 undergoing elective termination, 23 (55%) had syndromes or comorbidities vs. 280 (25%) of the 1135 live-births, $p = .0003$. Conclusions: In Nevada, despite a statistically significant increase in prenatal detection of significant congenital heart disease over time, termination of pregnancy rates did not increase. Nevertheless, those undergoing elective termination were more likely to have associated syndromes or comorbidities.},
doi={10.1080/14767058.2021.2004115},
url={https://doi.org/10.1080/14767058.2021.2004115}}

APÊNDICE I - ANÁLISE PELO SCOPUS DOS 577 RESULTADOS ENCONTRADOS PELOS DESCRITORES

Análise dos 577 resultados encontrados pelos descritores utilizados, realizada por meio da plataforma Scopus.



APÊNDICE J - ANÁLISE PELO WEB OF SCIENCE DOS 418 RESULTADOS ENCONTRADOS PELOS DESCRITORES

Análise dos 418 resultados encontrados pelos descritores utilizados, realizada pelo Web of Science.



APÊNDICE K - QUESTIONÁRIO DO GRUPO DE ESTUDO KOVACEVIC (2018)

1. Transfer of Medical Knowledge (sum-score range = 5 to 25; $\alpha = 0.798$, good)

1. I received sufficient medical knowledge concerning my child's heart defect.
2. I received the proper amount of medical information.
3. I am convinced the physician's explanation included all necessary details concerning my child's condition.
4. The possible consequences of my child's treatment were adequately explained to me.
5. Possible complications occurring during the treatment were explained well to me.

2. Transparency regarding the Treatment Process (sum-score range = 4 to 20; $\alpha = 0.808$, very good)

1. After counseling, I knew what would be the next steps in my child's treatment after delivery.
2. It was explained to me in an understandable way when and in what order the following steps in my child's treatment would take place.
3. It was explained to me in an understandable way why the following steps in my child's treatment would take place.
4. During the conversation, my questions were adequately answered.

3. Trust in Medical Staff (sum-score range = 3 to 15; $\alpha = 0.811$, very good)

1. Counseling has strengthened my trust in the medical institution.
2. The conversation strengthened my trust in the physician.
3. If possible, I would prefer that the same physician takes care of my baby after delivery.

4. Perceived Situational Control (sum-score range = 1 to 5, α not applicable as only one item)

1. During the conversation, I felt included in planning the treatment.

5. Coping Resources (sum-score range = 3 to 15, $\alpha = 0.743$, good)

1. I felt treated with proper compassion.
 2. The conversation helped me to cope with my concerns and fears.
 3. During the conversation, my questions and concerns were taken seriously.
-

Fonte: .Kovacevic, A.; et al 2022 <https://doi.org/10.3390/jcm11010278>

ANEXO A - REGISTRO NO OPEN SCIENCE FRAMEWORK (OSF)

The screenshot shows the OSFHOME interface. At the top, there is a navigation bar with 'OSFHOME' on the left and 'My Projects', 'Search', 'Support', 'Donate', and 'Marcela Bezerra Dias' on the right. Below this is a secondary navigation bar with 'Parental Counseling After Fetus Heart ...' selected, and other options like 'Files', 'Wiki', 'Analytics', 'Registrations', 'Contributors', and 'Settings'. The main content area displays the project title 'Parental Counseling After Fetus Heart Disease Diagnosis: a Scoping Review' with a file size of '121.1KB', a 'Public' status, and a 'P 0' indicator. Below the title, it lists contributors: 'Marcela Bezerra Dias, Bruno Mori, Luciane Alves da Rocha Amorim'. It also shows the creation date '2021-03-13 10:13 PM' and the last update date '2022-09-04 01:09 AM'. The category is 'Project'. A description follows: 'Counseling parents with a diagnosis of fetal heart disease can reduce issues and assist in the postnatal follow-up of this baby. This research proposes to map the evidence available in the literature on counseling parents diagnosed with fetal heart disease.'

ANEXO B - CONFIRMAÇÃO DE ARTIGO SUBMETIDO

← Submissions Being Processed for Author ①

Page: 1 of 1 (1 total submissions)

Results per page 10 ▾

Action 	Manuscript Number 	Title 	Initial Date Submitted 	Status Date 	Current Status 
View Submission Send E-mail		FAMILY COUNSELING AFTER THE DIAGNOSIS OF CONGENITAL HEART DISEASE IN THE FETUS: SCOPING REVIEW	02-23-2023	02-23-2023	Received

Page: 1 of 1 (1 total submissions)

Results per page 10 ▾

ANEXO C - CONFIRMAÇÃO DE ARTIGO APROVADO

De: Roseli Nomura <onbehalfof@manuscriptcentral.com>

Enviada em: quinta-feira, 23 de fevereiro de 2023 19:28

Para: araujojred@terra.com.br

Assunto: Revista da Associação Médica Brasileira - Decision on Manuscript ID RAMB-2023-0161

23-Feb-2023

Dear Prof. Araujo Júnior:

It is a pleasure to accept your manuscript entitled "What is important in family counseling in cases of fetuses with congenital heart disease?" in its current form for publication in the Revista da Associação Médica Brasileira.

Thank you for your fine contribution. On behalf of the Editors of the Revista da Associação Médica Brasileira, we look forward to your continued contributions to the Journal.

Sincerely,

Dr. Roseli Nomura

Editor-in-Chief, Revista da Associação Médica Brasileira

roseli.nomura@hotmail.com

ANEXO D - TABELA DE CONTRARREFERÊNCIA DOS ARTIGOS

Título do artigo	Contrarreferência
<p>1. Assessment of Needs for Counseling After Prenatal Diagnosis of Congenital Heart Disease - A Multidisciplinary Approach</p> <p>Kovacevic, A; et al.</p> <p>Alemanha</p> <p>2018</p>	<p>[1] Allan L, Dangel J, Fesslova V et al. Association for European Paediatric Cardiology. <i>Cardiol Young</i> 2004; 14: 109–1014 1-Estudos e exames de Prevenção</p> <p>[2] Allan LD, Huggon IC. Counselling following a diagnosis of congenital heart disease. <i>Prenat Diagn</i> 2004; 24: 1136–1142 Artigo de obs. em artigo consta em nosso banco excluídos 8 -Artigo de revisão relacionado ao tema</p> <p>[3] Arya B, Glickstein JS, Levasseur SM et al. Parents of children with congenital heart disease prefer more information than cardiologists provide. <i>Congenit Heart Dis</i> 2013; 8: 78–85 1 Artigo incluído na pesquisa.</p> <p>[4] Bertsche T, Neiningner MP, Kaune A et al. Interdisciplinary Concepts of Paediatrics and Clinical Pharmacy to Optimise Anticonvulsant Treatment. <i>Klin Padiatr</i> 2018; 230: 5–12 11- Fora do tema de pesquisa</p> <p>[5] Bonnet D, Coltri A, Butera G et al. Detection of transposition of the great arteries in fetuses reduces neonatal morbidity and mortality. <i>Circulation</i> 1999; 99: 916–918 11- Fora do tema de pesquisa</p> <p>[6] Bratt EL, Järholm S, Ekman-Joelsson BM et al. Parent's experiences of counselling and their need for support following a prenatal diagnosis of congenital heart disease – a qualitative study in a Swedish context. <i>BMC Pregnancy Childbirth</i> 2015; 15: 15 171 Artigo incluído na pesquisa.</p> <p>[7] Donofrio MT, Moon-Grady AJ, Hornberger LK et al. Diagnosis and treatment of fetal cardiac disease: a scientific statement from the American Heart Association. <i>Circulation</i> 2014; 129: 2183–2242 8 -Artigo de revisão relacionado ao tema</p> <p>[8] Franklin O, Burch M, Manning N et al. Prenatal diagnosis of coarctation of the aorta improves survival and reduces morbidity. <i>Heart</i> 2002; 87: 67–69 9- Artigo de revisão não relacionado ao tema</p> <p>[9] Gardiner HM, Kovacevic A, van der Heijden LB et al. Prenatal screening for major congenital heart disease: assessing performance by combining national cardiac audit with maternity data. <i>Heart</i> 2014; 100: 375–382 9- Artigo de revisão não relacionado ao tema</p> <p>[10] Holland BJ, Myers JA, Woods CR Jr. Prenatal diagnosis of critical congenital heart disease reduces risk of death from cardiovascular compromise prior to planned neonatal cardiac surgery: a meta-analysis. <i>Ultrasound Obstet Gynecol</i> 2015; 45: 631–638 9- Artigo de revisão não relacionado ao tema</p> <p>[11] Hotopp LC, Spindler UP, Bach VA et al. How do Parents Perceive the Initial Medical Consultation on their Child's Developmental Disorder? <i>Klin Padiatr</i> 2018; 230: 44–49 11 Fora do tema de pesquisa</p>

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