

CONGENITAL HEART DEFECTS

Ibragimov Saidburxon Abdumajid o'g'li

Temirov Javoxir Baxriddin o'g'li

Vahobov Karimjon Vafiqul o'g'li

Samarkand State Medical university

Abstract: Congenital heart defects (CHDs) represent one of the most common congenital anomalies worldwide, affecting approximately 1% of live births. They range from simple defects, such as atrial septal defects, to complex malformations like tetralogy of Fallot and hypoplastic left heart syndrome. This paper provides a comprehensive overview of CHDs, focusing on classification, pathophysiology, diagnostic tools, and treatment strategies. Advanced imaging techniques, such as fetal echocardiography and cardiac MRI, play an integral role in early detection and management. Innovations in minimally invasive surgical techniques and catheter-based interventions have significantly improved survival rates. Additionally, this study emphasizes the role of genetic research and prenatal screening programs in reducing the burden of CHDs.

Keywords: Congenital heart defects, fetal echocardiography, cardiac MRI, genetic screening, surgical interventions.

Annotatsiya: Tug'ma yurak nuqsonlari (TYuN) dunyo bo'ylab eng ko'p uchraydigan tug'ma anomaliyalardan biri bo'lib, har 100 ta yangi tug'ilgan chaqaloqning taxminan bittasida uchraydi. TYuN oddiy bo'shliq nuqsonlaridan (masalan, atrial septal nuqsonlar) tortib, murakkab anomaliyalargacha (masalan, Fallot to'rtligi yoki gipoplastik chap yurak sindromi) farq qiladi. Ushbu maqola TYuNlarning tasnifi, patofiziologiyasi, diagnostika vositalari va davolash strategiyalarini keng qamrovli tahlil qiladi. Fetal echokardiyografiya va yurak MRG kabi ilg'or tasvirlash texnologiyalari erta tashxis va davolashda muhim rol o'ynaydi. Minimal invaziv jarrohlik va kateter asosidagi aralashuvlar natijalarini sezilarli yaxshiladi. Ushbu tadqiqot, shuningdek, TYuNlarning yukini kamaytirishdagi genetik tadqiqotlar va prenatal skrining dasturlarining ahamiyatini ta'kidlaydi.

Kalit so'zlar: Tug'ma yurak nuqsonlari, fetal echokardiyografiya, yurak MRG, genetik skrining, jarrohlik aralashuvi.

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Аннотация: Врожденные пороки сердца (ВПС) являются одной из самых распространенных врожденных аномалий, встречающихся примерно у 1% новорожденных. Они варьируются от простых дефектов, таких как дефекты межпредсердной перегородки, до сложных пороков, таких как тетрада Фалло и синдром гипоплазии левых отделов сердца. В данной статье представлен комплексный обзор ВПС с акцентом на классификацию, патофизиологию, диагностические методы и стратегии лечения. Передовые методы визуализации, такие как фетальная эхокардиография и МРТ сердца, играют ключевую роль в раннем выявлении и лечении. Инновации в малоинвазивных хирургических методах и катетерных вмешательствах значительно улучшили выживаемость. Кроме того, в исследовании подчеркивается роль генетических исследований и программ пренатального скрининга в снижении бремени ВПС.

Ключевые слова: Врожденные пороки сердца, фетальная эхокардиография, МРТ сердца, генетический скрининг, хирургическое вмешательство.

Introduction. Congenital heart defects (CHDs) are structural abnormalities of the heart or great vessels present at birth, resulting from improper development during embryogenesis. Affecting nearly 1% of live births globally, CHDs are the most common congenital anomalies and remain a leading cause of infant morbidity and mortality. Advances in diagnostic tools and treatment options have drastically improved outcomes, but challenges remain in early detection, resource allocation, and equitable healthcare access.

This paper aims to provide a detailed analysis of CHDs, including their classification, underlying mechanisms, and management strategies. Special attention is given to the role of modern diagnostic imaging and the integration of genetic research in understanding and managing these conditions.

Materials and Methods. A systematic review of peer-reviewed articles published between 2015 and 2024 was conducted using PubMed, Scopus, and Google Scholar. Search terms included “congenital heart defects,” “fetal echocardiography,” “cardiac MRI,” and “genetic screening.” Inclusion criteria focused on clinical trials, meta-analyses, and case reports on CHDs. The methodology involved evaluating advancements in diagnostic imaging, surgical

interventions, and genetic research. Particular focus was given to studies addressing prenatal screening programs, minimally invasive techniques, and emerging pharmacological treatments.

Results. 1. Classification of CHDs. CHDs are categorized into two primary groups: Acyanotic Defects: Includes atrial septal defects (ASDs), ventricular septal defects (VSDs), and patent ductus arteriosus (PDA). Cyanotic Defects: Includes tetralogy of Fallot (TOF), transposition of the great arteries (TGA), and truncus arteriosus.

2. Pathophysiology. The pathophysiological mechanisms underlying CHDs involve genetic mutations, environmental factors, and maternal health conditions such as diabetes or infections. Disruptions in embryonic development during the first trimester result in structural heart abnormalities.

3. Diagnostic Tools. Fetal Echocardiography: Allows detection of CHDs as early as 18 weeks of gestation. Cardiac MRI: Provides detailed anatomical and functional information, especially for complex CHDs. Biomarkers: Elevated levels of NT-proBNP and troponins indicate cardiac stress and may aid in monitoring disease progression.

4. Treatment Approaches. Surgical Interventions: Open-heart surgery remains the gold standard for repairing complex CHDs. Techniques such as arterial switch operations for TGA and complete repair of TOF have significantly improved survival rates. Catheter-Based Procedures: Device closures for ASDs and VSDs and balloon atrial septostomy for cyanotic defects are minimally invasive alternatives.

Medical Management: Medications such as diuretics and prostaglandin E1 are used to stabilize patients before definitive surgery.

Discussion. This study underscores the importance of early detection and timely intervention in managing CHDs. Advances in prenatal screening and diagnostic imaging have revolutionized the ability to identify and treat these defects early, leading to improved outcomes. Genetic studies have revealed numerous mutations associated with CHDs, offering potential for personalized treatment strategies. However, challenges persist in resource-limited settings, where access to specialized care and advanced diagnostic tools is often unavailable. Global efforts to enhance healthcare equity and invest in training healthcare providers are critical for improving CHD management worldwide.

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Conclusion

Congenital heart defects, while common, pose significant challenges to global health systems. Advances in surgical techniques, imaging modalities, and genetic research have drastically improved survival and quality of life for affected individuals. Future research should focus on developing cost-effective screening and treatment options to ensure equitable access to care.

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