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CONTEMPORARY DIAGNOSTIC APPROACHES TO CONGENITAL HEART DEFECTS IN CHILDREN

Sheraliyeva R. M
Student of the Faculty of General Medicine,
Tashkent State Medical University, Tashkent, Uzbekistan,
e-mail: sheraliyevarobiya03@gmail.com

Alisherova M. A
Assistant of the Department of Medical Radiology,
Tashkent Medical Academy, Tashkent, Uzbekistan
e-mail: mahliyoalisherova1994@gmail.com

Abstract

Congenital heart defects are among the most common congenital cardiac anomalies observed in newborns and children. Their development is influenced by genetic, epigenetic, and environmental factors. Modern studies enable early detection of these defects through prenatal diagnostics and fetal echocardiography, which plays a crucial role in determining appropriate treatment strategies. Genetic and epigenetic research has demonstrated the impact of mechanisms such as chromosomal copy number variations, gene mutations, and DNA methylation on the development of congenital heart defects. Additionally, environmental factors—including maternal hypertension and exposure to heavy metals or pollution—also increase the risk. Clinical diagnosis is carried out using electrocardiography (ECG), echocardiography, pulse oximetry, and genetic testing, allowing for the development of individualized

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treatment plans for each patient. Furthermore, studies also examine the effects of these conditions on growth, development, and quality of life in affected children.

Keywords: Congenital heart defect (CHD), atrial septal defect (ASD), ventricular septal defect (VSD), Tetralogy of Fallot (TOF), prenatal diagnosis, fetal echocardiography, genetic factors, epigenetic changes, surgical intervention, pulse oximetry.

Introduction

Congenital heart defects (CHDs) are the most common type of congenital malformations. Among full-term newborns, CHDs are detected in approximately 8–10 per 1000 infants (0.8–1%) , while in preterm infants, this rate can increase tenfold, reaching up to 8.3% . The likelihood of CHDs is even higher during the early stages of fetal development, as some complex forms may lead to fetal demise (1). Surgical intervention is required in 50–60% of children born with CHDs, with 25% of these cases being critical . Moreover, CHDs are a leading cause of neonatal mortality .Early detection through ultrasound screening significantly improves treatment outcomes for many congenital heart conditions, allowing for planned delivery, genetic counseling, and appropriate perinatal monitoring (1). CHDs are among the most prevalent congenital disorders in children, arising from disruptions in the embryonic development of the heart and major blood vessels . These conditions can manifest with a wide range of clinical severity, from mild forms to life-threatening cases .The development of CHDs is influenced not only by genetic factors but also by environmental exposures (4). The resulting impairment of cardiac pump function may lead to heart failure, which clinically presents as respiratory difficulties, feeding problems, and delayed physical development in children, requiring timely diagnosis and treatment (6).The nine most common types of CHDs account for 80% of all cases, although prevalence rates vary across studies. These include Tetralogy of Fallot

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(4%) , pulmonary artery stenosis (9%) , atrial septal defect (5%) , patent ductus arteriosus (9%), aortic stenosis (5%), transposition of the great arteries (5%), and ventricular septal defect (36%). The remaining 20% consist of rare or complex defects.Early identification and timely treatment of suspected CHDs not only improve clinical outcomes but also reduce the risk of long-term complications (14).

Objective

The main aim of this article is to enable early detection of congenital heart defects and to study their developmental mechanisms.

Pathophysiology and Molecular Mechanisms

Congenital heart defects (CHD) are classified as mild, moderate, or severe depending on their primary anatomical structure and impact on the circulatory system . The most common types of CHDs include patent foramen ovale, atrial septal defect, ventricular septal defect, and patent ductus arteriosus, which together account for the majority of CHD cases .The heart is one of the earliest organs to develop during the embryonic period, and its formation is a tightly regulated process. Genetic-level changes can affect cell proliferation, differentiation, and migration, which are critical for proper embryonic development. Disruptions in genes encoding transcription factors, signaling pathways, or chromatin modifiers can halt proper cell differentiation and impair normal tissue and organ development, resulting in congenital malformations, including CHD (5).

The embryonic heart tube forms the atrioventricular (AV) canal, which separates the atrial and ventricular chambers and regulates the development of atrioventricular conductive tissue and valves. The AV canal is essential for the formation of the chambers and valves, and it also gives rise to pacemaker tissues that conduct impulses slowly from the atria to the ventricles. As the tube

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elongates, certain regions differentiate and proliferate to form the atria and ventricles, while the region between the atrial and ventricular chambers does not expand and remains as the AV canal. Consequently, during development, embryonic myocardial cells make a crucial decision: they either become AV canal pacemaker cells or working cardiomyocytes of the atrial and ventricular chambers(16).

The causes of congenital heart defects (CHD) are complex, and in approximately half of the cases, their pathogenesis remains unclear. At the same time, several genetic and environmental factors play a significant role in CHD development. For example, smoking is considered a teratogen, which can lead to morphological and functional abnormalities in the fetal cardiovascular system. Carbon monoxide and nicotine also negatively affect fetal heart development. Comprehensive meta-analyses have shown that maternal active or passive smoking, as well as paternal active smoking, increase the risk of CHD in children. Therefore, cessation of smoking by both parents before and during pregnancy is crucial for reducing the risk of CHD. Studies indicate that the risk of atrial septal defects and obstruction of the right ventricular outflow tract is particularly associated with maternal active smoking (5).

Types of Congenital Heart Defects and Clinical Manifestations

Atrial septal defect (ASD) is a congenital heart defect that arises from abnormal development of the atrial septum during the embryonic period and accounts for approximately 10–15% of patients with CHDs. In many cases, it remains asymptomatic until adulthood and may be detected incidentally . However, if the defect is large, patients may experience nonspecific symptoms such as shortness of breath, fatigue during physical activity, exercise intolerance, or frequent heart palpitations (5).

Atrial septal defects (ASD) vary depending on their location and morphology . In the majority of patients—over 80%—the secundum type, located in the region of

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the fossa ovalis, is observed and often presents with multiple defects. The size of the defects can vary; in some cases, they exceed 30 mm, or the primum atrial septum may be completely absent. This type of ASD can enlarge as the patient grows . Additionally, other types exist, including defects of the primum atrial septum (approximately 15%), defects in the venous inflow tract (5%, including the superior and inferior vena cava), and coronary sinus defects (<1%). ASDs can also occur in complex congenital heart defects, such as Tetralogy of Fallot or Ebstein anomaly (5).

Congenital heart defects (CHD) manifest in various forms, including atrial and ventricular septal defects, abnormalities of the arteries and ventricular outflow tracts, obstructive lesions in the left or right heart, and congenital anomalous venous connections. Ventricular septal defects (VSD) and atrial septal defects (ASD) are the most common types of CHDs worldwide . Defects of the ventricular outflow tracts, such as Tetralogy of Fallot, are associated with complex cyanotic manifestations. Obstructive lesions in the left or right heart, such as aortic stenosis or pulmonary stenosis, can lead to heart failure if left untreated. Anomalous venous connections may remain asymptomatic or cause cyanosis depending on the severity and type of the defect (5).

Patients with single-ventricle heart defects may exhibit symptoms such as fatigue, cyanosis (bluish discoloration of the skin and mucous membranes), rapid breathing, and difficulty feeding, which are related to inadequate pulmonary and systemic blood circulation.

Single-ventricle heart defects include:

- Left ventricle with a double inlet.
- Right ventricle with a double outlet and pulmonary atresia.
- Hypoplastic heart syndrome.
- Pulmonary atresia.
- Unbalanced congenital defects of the atrial and ventricular septa.

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Surgical correction is often performed in stages, using conduits or grafts to restore proper blood flow.(2)

Currently, the largest group of adults with congenital heart defects (CHDs) consists of those who underwent various treatments during childhood . This has significantly altered the presentation of CHDs in adults and requires cardiologists to be prepared to recognize new disease patterns. According to data from La Paz Hospital, 56% of adults under 40 belong to this group .The most common type of CHD is Tetralogy of Fallot, which was previously rare in general cardiology clinics but is now widely seen in its post-surgical form. Additionally, rare conditions such as transposition of the great arteries are also observed in clinical practice . Complete transposition is life-threatening if not surgically corrected during the first months of life. Today, many children reach adulthood through various surgical interventions, although their cardiac anatomy and function can change in diverse ways (3).

Tetralogy of Fallot develops during fetal life due to abnormal rotation of the cardiac outflow tracts, resulting in leftward and upward displacement of the infundibular (outflow) region . As a consequence, right ventricular outflow tract obstruction occurs, a ventricular septal defect forms due to malalignment of the septum, the aorta becomes overriding, and the right ventricle undergoes hypertrophy due to increased workload .The pulmonary valves are often small or stenotic, and the branches of the pulmonary artery may be hypoplastic, sometimes accompanied by distinct stenoses. In severe cases, both the right ventricular outflow tract and the pulmonary trunk may present with atresia.Additionally, approximately 25% of patients have a right-sided aortic arch, about 2% present with an atrial septal defect (commonly referred to as pentalogy of Fallot), and in rare cases, an atrioventricular canal defect is observed. Congenital anomalies of the coronary arteries occur in about 5% of patients and may play an important role in surgical correction.(25)

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Prenatal and Early Diagnosis

Ultrasound examination of the fetal heart allows detection of the majority of congenital heart diseases. However, when prenatal screening relies only on the four-chamber view, many conotruncal and outflow tract defects may remain undetected. Adding outflow tract assessment to the four-chamber view significantly improves the accuracy of ultrasound screening.In current practice, to enhance the detection of congenital heart defects, the standard four-chamber and outflow tract views are supplemented with the three-vessel view and the three-vessel–trachea view. In particular, the three-vessel–trachea view enables identification of anomalies such as aortic coarctation, right aortic arch, double aortic arch, and vascular rings. This approach significantly increases the sensitivity of prenatal diagnosis.(1)

Chromosomal microarray analysis (CMA) is recognized as an effective tool for detecting clinically significant copy number variations during pregnancy. Compared to karyotyping, CMA provides higher resolution for whole-genome screening and improves the detection of chromosomal imbalances and submicroscopic genetic alterations in both prenatal and postnatal periods. Therefore, CMA is expected to become a standard method for evaluating pregnancies at risk of congenital anomalies in the near future. It also supports informed decision-making for both clinicians and parents, although ethical considerations must be carefully addressed.In addition, variants of uncertain clinical significance present challenges in prenatal genetic counseling, as their impact and clinical outcomes remain unclear. Such variants may be identified in a considerable proportion of patients; for example, studies on hypertrophic cardiomyopathy have reported rates of up to 15%. The frequency of detecting these variants varies depending on the number and characteristics of the genes being analyzed (22).

Congenital heart defects represent a significant issue in pediatrics due to their high prevalence, potential to cause serious health complications and disability in

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children, and the frequent need for early surgical intervention. According to the World Health Organization, these defects occur in approximately 0.7–1.7% of newborns. Understanding the underlying mechanisms of these pathological conditions and utilizing modern research methods for their early detection enable timely diagnosis and, when necessary, appropriate correction of developmental abnormalities in children (12).

Congenital heart defects (CHD) are often associated with the right ventricle and the tricuspid valve. During pregnancy, fetal ultrasound and echocardiography can detect structural abnormalities and disturbances in blood flow. In newborns, signs such as murmurs, cyanosis, or difficulty breathing are identified through screening, with simple tests like pulse oximetry playing a key role in the early detection of critical CHD. After birth, patients undergo detailed evaluation using a stethoscope, ECG, X-ray, and echocardiography, which are essential for assessing heart structure and right ventricular function (8).In some cases, CHD may have a genetic or syndromic component, warranting the use of karyotyping and genomic testing. Genetic analysis helps determine the underlying causes of the disease and guides treatment planning (9). Early diagnosis improves growth, development, and surgical outcomes in children, and systematic screening enhances the timely identification of the condition (7).

Epidemiological data play a crucial role in assessing the risk of congenital heart defects (CHD), which occur in approximately 0.7–1.7% of newborns. The detection process involves several stages, beginning with the evaluation of the patient's clinical condition, such as difficulty breathing, fatigue, or changes in skin color. Subsequently, the heart rhythm and structure are assessed using ECG and echocardiography (11,15). In newborns, pulse oximetry is used to determine blood oxygen saturation. During pregnancy, fetal echocardiography can identify certain heart defects in advance (13). When necessary, genetic testing is performed to detect chromosomal or genetic mutations. These methods enable

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early diagnosis of congenital heart defects and help guide the patient's subsequent treatment plan (10,17).

The diagnosis of congenital heart defects (CHD) takes into account not only clinical signs but also genetic and environmental factors. Studies have shown that exposure to harmful dust particles in the maternal environment can increase the risk of CHD (18). Clinical evaluation involves assessing heart rhythm, structure, and blood flow using ECG and echocardiography, along with monitoring oxygen saturation. Genetic diagnostics help identify copy number variations or anomalies in cell division (19). Additionally, genetic testing can detect disruptions in cell division and the boundaries of heart compartments, which is important for understanding the developmental mechanisms of CHD (20). Moreover, exposure of the mother and infant to metals, such as cobalt, is also considered in the assessment. By analyzing clinical, genetic, and environmental factors together, CHD can be detected at an early stage, and an appropriate treatment strategy can be determined (21).

The detection of congenital heart defects (CHD) is carried out in several stages. Initially, clinical examinations assess the infant's breathing difficulties, fatigue, changes in skin color, and heart murmurs (24). In newborns, ECG and echocardiography are used to evaluate heart rhythm and structure, while pulse oximetry measures oxygen saturation. Genetic testing helps identify chromosomal and genetic mutations, and the study of DNA methylation and other epigenetic changes provides insight into the developmental mechanisms of CHD (23). Clinical and genetic findings are combined to confirm the diagnosis. During pregnancy, fetal echocardiography can allow early detection of certain defects. Diagnostic results guide the timing and planning of surgical interventions, as early or delayed procedures can affect patient outcomes. In this way, clinical, genetic, and epigenetic data are integrated to enable early detection of CHD and the development of individualized treatment strategies (24).

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Discussion

Congenital heart defects (CHD) are widespread among both children and adults, with their development arising from a complex interaction of genetic, epigenetic, and environmental factors (5). Genetic mutations, chromosomal imbalances, and disruptions in GATA transcription factors can interrupt cardiac cell differentiation, leading to septal and conotruncal defects. Additionally, epigenetic mechanisms such as DNA methylation play a significant role in the pathogenesis of CHD (23). Environmental influences—including maternal smoking, pollution, and exposure to metals—adversely affect fetal heart development and increase the risk of CHD (21).

Prenatal screening, including fetal echocardiography and chromosomal microarray analysis, enables early detection of CHD, which helps optimize surgical planning and treatment strategies (1, 22, 24). Severe defects present with symptoms such as shortness of breath, fatigue, and heart failure, and complex conditions like Tetralogy of Fallot or transposition of the great arteries require surgical intervention (2, 24). Therefore, designing individualized treatment plans and optimizing surgical timing for patients with CHD can significantly improve quality of life (25). In conclusion, considering clinical, genetic, and environmental factors, prevention, early diagnosis, and personalized therapy play a crucial role in optimizing outcomes for CHD (5).

Conclusion

Congenital heart defects (CHD) arise from the complex interplay of genetic, epigenetic, and environmental factors. These conditions manifest as ventricular and atrial defects, conotruncal anomalies, and malformations of the cardiac outflow tracts. Maternal smoking, exposure to particulate matter, and metals increase the risk of CHD development. Early detection of CHD is possible through prenatal ultrasound, three-vessel and trachea views, chromosomal microarray analysis (CMA), and postnatal screening. By integrating clinical and

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genetic data, individualized treatment plans can be developed, allowing for optimal timing of surgical interventions, improved quality of life, and better long-term prognosis. Furthermore, in adults with CHD, post-surgical anatomical changes and complex cardiac structures necessitate new diagnostic and therapeutic approaches. Therefore, early diagnosis and preventive measures play a crucial role in reducing the complex complications associated with CHD.

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