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REVIEW ARTICLE

A DETAILED REVIEW OF CONGENITAL HEART DISEASE IN INFANTS AND CHILDREN

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ABSTRACT
Congenital heart disease (CHD) comprises structural abnormalities of the heart or great vessels that develop before birth. Its presentation ranges from small defects that may require observation to critical conditions associated with cyanosis, shock or heart failure in the neonatal period. The supplied source document describes the developmental basis, major types, altered circulation, clinical features, diagnostic methods and management of CHD. Genetic and chromosomal factors may contribute, while maternal conditions and environmental exposures are also discussed; however, the cause of many cases remains unknown. Prenatal imaging, newborn examination and pulse-oximetry screening can support early recognition, while echocardiography is central to diagnosis. Management is individualized and may include monitoring, medicines for stabilization or complications, catheter-based intervention and surgery. As survival improves, long-term follow-up is important to identify residual lesions, arrhythmias, ventricular dysfunction and developmental or psychosocial needs. This review summarizes these topics and emphasizes early referral, safe medication use and coordinated care across childhood and adulthood.

Keywords: *congenital heart disease, infants, children, cyanosis, echocardiography, cardiac surgery, screening.*

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1. INTRODUCTION

Congenital heart disease (CHD) refers to abnormalities in the structure or function of the heart and major blood vessels that are present at birth. The defect develops during fetal life, but symptoms may become evident immediately after delivery or emerge months or years later. Some children have few symptoms, whereas severe lesions can compromise oxygen delivery or systemic blood flow and become medical emergencies.

The supplied document reports that CHD is among the most common congenital disorders and cites contemporary global estimates approaching 9–10 cases per 1,000 live births, with variation between regions and studies. Increased detection of milder lesions contributes to differences in reported prevalence. Advances in fetal echocardiography, newborn screening, neonatal intensive care, catheter-based procedures, and cardiac surgery have improved recognition and survival. Consequently, treatment now includes not only anatomical correction but also monitoring of heart function, growth, development, quality of life, and risks that may persist into adulthood [1].

1.1 Purpose and Scope

This article reviews the main themes of the source document, including causes and risk factors, embryology, classification, pathophysiology, clinical presentation, diagnosis, treatment, complications, prevention, recent advances, and lifelong follow-up. It is an educational review and does not replace assessment by a qualified healthcare professional.

2. REVIEW OF THE LITERATURE AND REVIEW MATTER

The supplied document brings together information on the epidemiology, developmental origins, clinical assessment, and treatment of CHD, supported by public health information, review articles, and clinical guidance listed in its references. Across these topics, a consistent theme is that CHD is not a single disease but a diverse group of lesions with different effects on circulation and different treatment needs. The cited literature also reflects a shift from focusing solely on survival after repair towards lifelong monitoring and patient-centred outcomes.

2.1 Etiology and Risk Factors

The heart develops early in pregnancy, particularly during approximately the third to eighth weeks. In many patients, no single cause can be identified; CHD is often considered multifactorial. The source identifies chromosomal conditions such as Down syndrome (trisomy 21), Turner syndrome (45,X), Edwards syndrome (trisomy 18), and Patau syndrome (trisomy 13), as well as selected single-gene changes, as possible genetic contributors.

Maternal and environmental factors described in the document include diabetes, obesity, rubella and other infections, poor nutritional status, folate deficiency, phenylketonuria, certain autoimmune diseases, alcohol consumption, smoking or tobacco exposure, ionizing radiation, industrial chemicals, and environmental pollutants. These are risk factors rather than proof of causation in an individual pregnancy. Preconception counselling, management of maternal conditions, and professional review of medicines and exposures can reduce some avoidable risks.

2.2 Embryology and Developmental Basis

Normal cardiac formation involves the development and looping of the primitive heart tube, formation of chambers and valves, septation, and alignment of the great arteries. Incomplete or abnormal septation can result in an atrial septal defect (ASD) or ventricular septal defect (VSD). Abnormal outflow tract development can contribute to tetralogy of Fallot, truncus arteriosus, and aortic arch abnormalities.

Fetal circulation also differs from postnatal circulation because the placenta provides oxygen and fetal shunts bypass much of the lungs. After birth, pulmonary resistance falls and systemic resistance rises. In selected duct-dependent defects, closure of the ductus arteriosus can cause rapid deterioration.

2.3 Classification

Clinically, CHD is classified according to cyanosis, direction of shunting, pulmonary blood flow, anatomy, and severity. Acyanotic lesions commonly include ASD, VSD, atrioventricular septal defect (AVSD), and patent ductus arteriosus (PDA), which may produce left-to-right shunting and increased pulmonary blood flow.

Cyanotic lesions include tetralogy of Fallot, pulmonary atresia, tricuspid atresia, transposition of the great arteries, total anomalous pulmonary venous return, and truncus arteriosus. Some complex defects have variable physiology, so classification must be interpreted alongside detailed cardiac imaging [2].

Table 01: Classification of Congenital Heart Disease

Classification basis	Examples or defining features
Cyanosis	Acyanotic lesions versus lesions causing low systemic oxygen saturation
Shunt direction	Left-to-right: ASD, VSD, PDA; right-to-left: examples include tetralogy of Fallot
Pulmonary flow	Increased, decreased, or variable, depending on the defect and associated obstruction
Anatomy/severity	Septal or valve defects; complex or critical lesions requiring urgent care

3. PATHOPHYSIOLOGY

The physiological effect of a congenital defect depends on its size, location, and associated abnormalities. A septal opening can permit abnormal shunting; a narrowed or absent valve can obstruct blood flow; and an abnormal connection can cause oxygenated and deoxygenated blood to mix or circulate through parallel pathways. These changes alter pressure, oxygen saturation, and the amount of blood reaching the lungs and body. In left-to-right shunts, oxygenated blood returns to the right side of the heart and is sent back to the lungs. Significant excess pulmonary blood flow can enlarge cardiac chambers and, over time, contribute to pulmonary hypertension. In right-to-left shunts, deoxygenated blood enters the systemic circulation and lowers arterial oxygen saturation, producing cyanosis. Pressure or volume overload can lead to hypertrophy, dilation, reduced cardiac output, and heart failure. Large untreated shunts may, in some cases, cause irreversible pulmonary vascular changes and Eisenmenger physiology.

4. CLINICAL MANIFESTATIONS

Symptoms vary depending on the lesion and the child's age. Newborns with critical disease may develop cyanosis, rapid breathing, poor feeding, lethargy, poor perfusion, or shock. Infants with significant shunts may sweat or breathe rapidly during feeding, gain weight poorly, or experience recurrent respiratory symptoms. Older children may show exercise intolerance, a heart murmur, or growth concerns. Adolescents and adults may experience palpitations, fainting, breathlessness, or reduced exercise capacity. Some defects are first suspected following the detection of an incidental murmur or an abnormal test result. Clinical examination may include assessment of oxygen saturation, respiratory and heart rates, blood pressure, peripheral pulses, heart sounds, murmurs, liver enlargement, and signs of heart failure. Weak femoral pulses or a difference between upper- and lower-limb blood pressure may suggest coarctation of the aorta. No symptom or murmur alone confirms CHD. Cyanosis, respiratory distress, poor perfusion, or shock requires urgent medical evaluation [3].

5. DIAGNOSIS AND SCREENING

Prenatal ultrasound can identify some structural abnormalities. Fetal echocardiography provides a more detailed assessment when screening is abnormal or risk is increased and may guide delivery planning at a centre with appropriate neonatal and cardiac services.

After birth, physical examination and pulse oximetry screening can detect some critical defects, including conditions that are not obvious on routine examination. A normal screening result does not rule out every defect.

Table 02: Diagnostic Investigations for Congenital Heart Disease

Investigation	Main contribution	Important limitation
Echocardiography	Shows anatomy, valves, chamber size, function, and Doppler blood flow	Image quality can limit detail
ECG	Assesses rhythm, conduction, and possible chamber enlargement	Does not define anatomy by itself
Chest X-ray	May show heart size and pulmonary vascular pattern	Limited specificity
Cardiac MRI/CT	Provides detailed anatomy; MRI can measure volumes and flow, while CT shows vessels at high resolution	Availability and patient-specific limitations; CT uses ionizing radiation
Cardiac catheterization	Measures pressures and oxygen saturations; may permit device or balloon treatment	Invasive and used when clinically indicated

Echocardiography is the central diagnostic test for many lesions. The choice of additional tests depends on the suspected defect and the information needed for treatment planning. Prompt referral is important when symptoms or screening suggest critical disease [4].

6. MANAGEMENT

Treatment is selected according to anatomy, physiology, age, symptoms, associated conditions, and specialist assessment. Some small defects can be observed with planned follow-up. Other patients need stabilization, catheter-based intervention, or surgery. The source emphasizes that medicines generally control symptoms or complications rather than correct the underlying structural defect.

6.1 Pharmacological Management

Prostaglandin E1 (alprostadil) may be used in selected newborns to keep the ductus arteriosus open when systemic or pulmonary blood flow depends on it, while definitive care is arranged.

Diuretics such as furosemide may relieve congestion in heart failure. ACE inhibitors or ARBs, beta-blockers, and digoxin may be used in selected clinical situations, depending on the lesion and the child's condition. Antiplatelet or anticoagulant therapy is reserved for specific indications, and pulmonary vasodilators may be used for selected patients with pulmonary arterial hypertension under specialist care.

Paediatric medication safety is essential because doses are often weight-based. The source highlights the importance of verifying the current weight, dose, concentration, route, frequency, and measuring devices, together with monitoring renal function, electrolytes, adverse effects, and adherence. Medicines should not delay the correction of a significant lesion when intervention is indicated.

6.2 Catheter-Based and Surgical Management

Interventional cardiology offers less invasive options for selected defects. Procedures include device closure of suitable ASDs or PDAs, balloon valvuloplasty for certain valve stenoses, and angioplasty or stenting for selected vascular obstructions.

Surgical procedures may include patch closure of septal defects, PDA ligation, repair of tetralogy of Fallot, reconstruction of an obstructed vessel or aortic arch, and complex neonatal or staged procedures for single-ventricle conditions. Decisions require a specialist team and depend on the individual anatomy.

7. COMPLICATIONS AND LONG-TERM CARE

Possible complications include heart failure, ventricular dysfunction, pulmonary hypertension, arrhythmias, residual shunts, valve regurgitation, recurrent obstruction, aortic complications in selected lesions, thromboembolism, and infective endocarditis in susceptible patients. Some children with complex CHD may experience feeding and growth problems or neurodevelopmental, learning, behavioural, and psychosocial difficulties. Risks differ according to the lesion, residual anatomy, ventricular function, genetic conditions, and previous procedures.

Repair in childhood does not always eliminate future risks. Lifelong or long-term congenital cardiology follow-up may be needed to monitor heart rhythm, ventricular function, exercise capacity, and late complications. Transition to adult

congenital heart services helps maintain continuity of care and supports individualized advice about lifestyle and reproductive health.

8. PREVENTION AND PUBLIC HEALTH

Not all CHD can be prevented because genetic and multifactorial mechanisms are not always modifiable. The source recommends preconception counselling where there is a personal or family history, good control of maternal diabetes, appropriate vaccination and rubella prevention before pregnancy, management of maternal metabolic disorders, and professional review of medicines and potentially harmful exposures. Genetic counselling and testing may be considered when a familial or syndromic cause is suspected.

Fetal echocardiography may be appropriate for pregnancies meeting high-risk criteria, while newborn pulse oximetry screening can support the early detection of critical CHD. Early referral is warranted when cyanosis, poor feeding, abnormal pulses, a concerning murmur, or signs of heart failure are identified.

Public health priorities include access to paediatric cardiology, echocardiography, catheterization, surgery, and long-term follow-up, especially in areas where diagnosis and specialist treatment are limited.

9. RECENT ADVANCES AND FUTURE DIRECTIONS

The source describes advances in prenatal imaging, three-dimensional echocardiography, cardiac MRI and CT, and the use of three-dimensional printing and virtual or augmented reality for selected planning and educational purposes. Catheter-based interventions continue to expand less invasive treatment options, while improvements in neonatal intensive care, anaesthesia, and surgical techniques have improved outcomes for complex disease [5]. Genomic testing can identify chromosomal abnormalities, copy-number changes, and selected single-gene causes. Such findings may support counselling and assessment for associated conditions, but variants require careful interpretation.

Future priorities include better long-term outcome data, more reliable risk prediction, improved access to specialized care, prevention of neurodevelopmental complications, and personalized surveillance.

10. CONCLUSION

Congenital heart disease is a broad group of abnormalities with outcomes ranging from mild, asymptomatic defects to life-threatening neonatal emergencies. Genetic, maternal, and environmental factors may contribute, while the cause of many cases remains unknown. Understanding embryology and the resulting changes in blood flow helps explain differences in symptoms and severity.

Prenatal assessment, newborn examination, pulse oximetry screening, and echocardiography support early diagnosis. Treatment may involve observation, medicines, catheter-based procedures, or surgery, with the choice tailored to the defect and the child. As survival improves, ongoing follow-up is important for identifying residual lesions, arrhythmias, ventricular dysfunction, and developmental needs. Coordinated, lifelong care aims to improve survival, function, and quality of life.

11. FINANCIAL SUPPORT

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12. DECLARATION COMPETING INTEREST

Not Declared

13. ACKNOWLEDGEMENT

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14. INFORM CONSENT AND ETHICAL CONSIDERATION

Not Applicable

15. REFERENCES

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