Congenital Heart Disease

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KEYWORDS

• Congenital heart disease • Pediatric cardiology • CHD

KEY POINTS

- Although congenital heart disease (CHD) may be detected prenatally with fetal echocardiogram and neonatally with pulse oximetry, some CHD lesions remain undetected until symptoms develop.
- CHD can have ongoing clinical implications throughout the life span. Children, adolescents, and adults living with CHD require ongoing care for medical issues specifically related to CHD.
- Care for children with CHD requires collaboration between the primary care provider and pediatric specialists.

INTRODUCTION

Congenital heart disease (CHD) accounts for almost one-third of all congenital birth defects and affects nearly 1 in 100 births annually in the United States.[1–3] Global prevalence of CHD has increased by 4.2% from 1990 to 2017.[1] Mortality related to CHD has decreased significantly due to advances in prenatal diagnosis, early intervention, and surgical correction.[1,3,4] Most children with CHD are now expected to reach adulthood.[3] Global estimates for people living with CHD have increased by 18.7% since 1990 with more than 2 million infants, children, adolescents, and adults living with CHD in the United States.[1,2] People living with CHD have complex medical needs and require their primary care physician and medical home to collaborate with specialists, promote care coordination, anticipate unique health needs, and address psychosocial issues for both the patient and family.[5,6]

Causes of Congenital Heart Disease

The exact cause of most CHD remains unknown apart from CHD results from changes early in development in utero. CHD is a multifactorial disease with both a genetic inheritance pattern, which typically involves more than a single gene, and influence

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from environmental factors.[7] Approximately 15% of CHD is thought to be associated with genetic conditions.[8] **Table 1** describes several genetic syndromes associated with CHD. Consultation with a pediatric geneticist may be warranted when there is concern for a genetic syndrome associated with CHD diagnosis. Some genetic syndromes are evaluated prenatally with cell-free fetal DNA testing and can warrant further evaluation with a fetal echocardiogram if needed.[9]

Risk factors associated with CHD development include[9,10] the following:

- Medications: retinoic acid, thalidomide, angiotensin-converting enzyme (ACE) inhibitors, statins, paroxetine, lithium, and sodium valproate
- Maternal infection with rubella during pregnancy
- Pregestational maternal diabetes, particularly when uncontrolled. Gestational diabetes does not seem to increase the risk of CHD development.
- Environmental exposures to organic solvents
- Maternal obesity or smoking

SCREENING

Prenatal Screening

Screening for CHD can begin as early as prenatally using fetal echocardiogram. Fetal echocardiogram includes comprehensive ultrasound views of cardiac structures as well as outflow tracts.[11] Fetal echocardiogram is not currently recommended as a universal screening test due to limited cost effectiveness but may be indicated when the risk of CHD exceeds the general population (typically >3%).[9] Maternal and fetal risk factors that would be appropriate for fetal echocardiogram include the following:[9,11,12]

Maternal indications

- Maternal pregestational diabetes mellitus
- Maternal phenylketonuria
- Maternal antibodies for anti-SSA (anti–Sjögren's-syndrome-related antigen A)/Ro and anti-SSB (anti–Sjögren's-syndrome-related antigen B)/La
- Maternal medication uses during pregnancy, which includes ACE inhibitors, retinoic acid, or nonsteroidal anti-inflammatory agents (specifically in third trimester)
- Maternal rubella infection during first trimester
- Maternal infection with concern for fetal myocarditis
- Conception with assisted reproduction technology
- CHD in first-degree relative
- First or second-degree relative with genetic disorder associated with CHD (22q11.2 deletion syndrome or DiGeorge syndrome, Noonan, CHARGE syndrome)

Fetal indications

- Fetal cardiac abnormality suspected on obstetric ultrasound
- Fetal major extracardiac anomaly suspected on obstetric ultrasound
- Fetal karyotype abnormality with positive cell-free fetal DNA testing or by diagnostic testing
- Persistent fetal tachycardia or bradycardia
- Persistent fetal arrhythmia (apart from isolated premature atrial contractions)
- Fetal neural tube measurement 3 mm or greater
- Monochorionic twin pregnancy
- Fetal hydrops

Table 1
Genetic syndromes associated with congenital heart disease

Genetic Syndrome	Chromosome Change	Features	Commonly Associated CHD
Patau syndrome	Trisomy 13	Microcephaly, cleft palate/lip, intellectual disability, polydactyly, omphalocele	ASD, VSD, PDA, HLHS
Edwards syndrome	Trisomy 18	Microcephaly, omphalocele, intellectual disability, intrauterine growth restriction, polyhydramnios, rocker bottom feet or clubfoot	ASD, VSD, PDA, TOF, TGA, CoA
Down syndrome	Trisomy 21	Mental impairment, stunted growth, umbilical hernia, hypotonia, epicanthal folds, upslanting palpebral fissures, single palmar transverse crease	ASD, VSD, TOF, PDA
Turner syndrome	Monosomy X	Webbed neck, short stature, low-set ears, diabetes, infertility, widely spaced eyes, growth issues	Bicuspid aortic valves, CoA
Wolf-Hirschhorn syndrome	Partial deletion on Chromosome 4 (4p16.3)	Microcephaly, micrognathia, short philtrum, intellectual disability, hypotonia, seizures	ASD, VDS, PDA
Cri-Du-Chat	Chromosome 5 deletion	Characteristic cry, feeding issues, mutism	VSD, ASD, PDA, TOF
DiGeorge syndrome	22q.11.2 Deletion syndrome	Characteristic facial features, palatal abnormalities, feeding issues, hypocalcemia, immune deficiency/thymus anomalies, learning difficulties, psychiatric disease	IAA, TA, TOF, VSD

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Table 1
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Genetic Syndrome	Chromosome Change	Features	Commonly Associated CHD
CHARGE syndrome	Mutation on CHD7 gene on Chromosome 8	Coloboma of eye, atresia of choanae, retardation of growth/ development, genital/urinary defects, ear anomalies/ deafness	TOF, PFA, AVSD, VSD
Williams syndrome	Deletion on Chromosome 7s	Unusual facial features, poor growth, hypotonia, intellectual disability, hypercalcemia	Supravalvular AS, PS, VDS, ASD

Abbreviations: ASD, atrial septal defect; CoA, coarctation of the aorta; HLHS, hypoplastic left heart syndrome; IAA, interrupted aortic arch; PDA, patent ductus arteriosus; TA, truncus arteriosus; TGA, transposition of the great arteries; TOF, Tetralogy of Fallot; VSD, ventricular septal defect.

Data source [Pierpont ME, Brueckner M, Chung WK, Garg V, Lacro RV, McGuire AL, Mital S, Priest JR, Pu WT, Roberts A, Ware SM, Gelb BD, Russell MW; American Heart Association Council on Cardiovascular Disease in the Young; Council on Cardiovascular and Stroke Nursing; and Council on Genomic and Precision Medicine. Genetic Basis for Congenital Heart Disease: Revisited: A Scientific Statement From the American Heart Association. Circulation. 2018 Nov 20;138(21):e653-e711. https://doi.org/10.1161/CIR.0000000000000606. Erratum in: Circulation. 2018 Nov 20;138 (21):e713. PMID: 30571578; PMCID: PMC6555769.][7]

The most commonly detected CHD by fetal echocardiogram is Tetralogy of Fallot (TOF), followed by transposition of the great arteries, followed by hypoplastic left heart syndrome, pulmonary atresia, total anomalous pulmonary venous return (TAPVR), tricuspid atresia, and truncus arteriosus to a lesser degree. Although fetal echocardiogram has greatly improved the detection of serious CHD, more than 50% of newborns with CHD are unrecognized at birth.[8,13,14]

Neonatal Screening

Screening for critical congenital heart defects (CCHD) aims to reduce the incidence of death and other poor health outcomes in newborns before the onset of symptoms. Screening for CCHD has also become an important tool at discriminating between CCHD and other diagnoses that present with shock, cyanosis, or respiratory distress. Screening with pulse oximetry testing (POS) has been shown to be effective at significantly reducing infant deaths due to CHD.[13,14] One observational study of more than 27 million births from 2007 to 2013 showed a decline of 33.4% in the death rate associated with POS.[14]

Screening for CCHD is a simple test to administer (**Fig. 1**). The recommendations were updated by the American Association of Pediatrics (AAP) in 2018, which stated pulse oximetry levels should be documented in both hand and foot. Oxygen levels would need to be measured at greater than or equal to 95% at both locations to pass screening. Additionally, the difference between arm and leg to pass screening must be less than or equal to 3% to be considered a normal screen. The AAP also

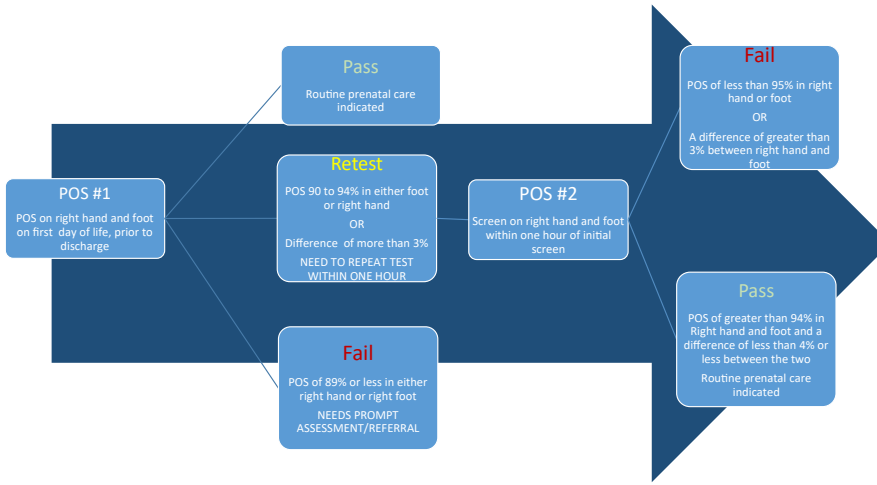

Fig. 1. Algorithm for CCHD screening with pulse oximetry. (Data source [Martin GR, Ewer AK, Gaviglio A, Hom LA, Saarinen A, Sontag M, Burns KM, Kemper AR, Oster ME. Updated Strategies for Pulse Oximetry Screening for Critical Congenital Heart Disease. Pediatrics. 2020 Jul;146(1):e20191650. https://doi.org/10.1542/peds.2019-1650. Epub 2020 Jun 4. PMID: 3249938].)

recommends a single repeat screen for newborns who do not pass screening initially.[15] Infants with a positive screen are recommended to undergo diagnostic echocardiogram.

These recommendations have been adopted by nearly all states in the United States (except for New Jersey and Tennessee, which have adopted similar strategies). Ideally, screening should occur before discharge on all infants born in the hospital, within the first day or 2 of life to identify newborns before presentation but also note that the false-positive rate is higher if screening is completed within the first 24 hours of life (0.47%–0.5% vs 0.05%–0.11%).[16]

Newborn screening improves outcomes after antenatal screening as well. Although antenatal ultrasound is a useful tool for detection of CCHDs, the sensitivity of prenatal ultrasound to detect certain abnormalities does vary for the detection of CCHD (**Table 2**).[17] Approximately a third are missed in the antenatal imaging, which could later be found on newborn POS. In a study that examined more than 700,000 newborns, the percentage of newborns testing positive on the screen has remained low with positive screening rates of 11.4 per 10,000 (95% CI 5.1–25.2) and the number of identified newborns with CCHD was 0.9 per 10,000 (95% CI 0.4–2.3).[16]

PRESENTATION

Fetal circulation depends on the placenta for gas exchange and oxygenation. Only 10% of blood is circulated to the fetal lungs. Two shunts are used in utero to assist with fetal circulation and bypassing of the lungs using high pulmonary vascular resistance. The foramen ovale allows shunting of fetal blood from the right atrium to the left atrium. The ductus arteriosus (DA) shunts blood from the right ventricle to the aorta connecting the trunk of the pulmonary artery to the proximal descending aorta using prostaglandins (PGE).

Table 2
Prenatal ultrasound sensitivity

Most Sensitive (>60%)	More Sensitive (25%–59%)	Least Sensitive (<25%)
Hypoplastic left heart	Tricuspid atresia	Ebstein anomaly
d-transposition of great arteries	Critical AS	TOF
Pulmonary atresia	Double-outlet right ventricle	Interrupted aortic arch
Truncus arteriosus		CoA
Critical PS		
Total anomalous pulmonary venous drainage		
Single ventricle		

Data source [Martin GR, Ewer AK, Gaviglio A, Hom LA, Saarinen A, Sontag M, Burns KM, Kemper AR, Oster ME. Updated Strategies for Pulse Oximetry Screening for Critical Congenital Heart Disease. Pediatrics. 2020 Jul;146(1):e20191650. https://doi.org/10.1542/peds.2019-1650. Epub 2020 Jun 4. PMID: 3249938].

Following delivery, the neonate uses the lungs for oxygenation of blood rather than the placenta. Increased arterial oxygen tensions in combination with a reduction in PGE and decreased pulmonary vascular resistance changes shunting left-to-right. The PDA is expected to close following delivery although some changes in neonatal circulation may continue for the first 6 to 8 weeks of life.[18,19]

Although there are methods to screen for CHD including fetal echocardiogram and POS following delivery, CHD may remain undiagnosed until signs and symptoms of CHD are detected. Signs and symptoms for nonspecific CHD may include tachypnea, irritability, cyanosis, pallor, weak peripheral pulses, or feeding intolerance that may be associated with poor weight gain.[18,19] Evaluation by the primary care provider during well child visits continues to be an important way to detect CHD.

CHD lesions can vary based on the timing of presentation. More severe lesions including TAPVR, hypoplastic left heart syndrome (HLHS) and dextro-transposition of the great arteries (D-TGA) requiring urgent evaluation for surgical intervention generally present directly following delivery. Many other cyanotic CHD lesions present within the first week to month of life as do ductal-dependent acyanotic lesions such as aortic stenosis (AS) and coarctation of the aorta (CoA). Acyanotic CHD with left-to-right shunt lesions may present beyond 1 month of life.[18–21]

Cyanotic Congenital Heart Disease

Cyanosis can be determined by physical examination and pulse oximetry screening. Most ductal-dependent lesions are cyanotic, and symptoms occur within hours to days of life. POS is particularly sensitive in detecting cyanotic CHD. Infants with lesions that require PDA support may present in shock in the first week of life so a high index of suspicion with a low threshold to start prostaglandin E_1 (PGE_1) in the first week of life is important for appropriate treatment of CHD.[22] If an infant presents with symptoms of cyanosis and a heart murmur, workup should include chest x-ray and echocardiogram with Doppler imaging interpreted by a pediatric cardiologist. If an echocardiogram is not available and there is a high probability of a ductal-dependent cardiac lesion, PGE should be started. A hyperoxia test can be used to help distinguish cyanotic CHD form pulmonary disease. Cyanotic CHD includes left heart obstructive lesions, right heart obstructive lesions, and mixing lesions (**Table 3**).

Table 3
Cyanotic congenital heart disease

	Presentation	Timing	Intervention
TAPVR	Cyanosis, respiratory distress and signs of decreased cardiac output	First few hours of life	Urgent surgery
D-TGA + restrictive ASDs +adequate blood mixing from ASD	Severe cyanosis and possibly acidosis • Possible increase in left ventricle impulse • Holosystolic murmur along left sternal border • S_2 is singular • Pulses in the lower extremity may be week or absent	At birth	PGE; may need urgent balloon septostomy to enlarge ASD
	Mild-to-moderate cyanosis at birth	At birth to first few days	PGE until definitive repair in the first week of life typically
Truncus arteriosus	• Murmur and minimal cyanosis • If left untreated, cyanosis results and heart failure	Newborn 1–2 mo old	Anticongestive medications Surgery
Hypoplastic left heart syndrome	Cyanosis, respiratory distress (especially with feeding) and signs of low cardiac output	Often diagnosed before birth, if not within hours or days before birth	PGE_1 until Norwood surgical procedure; may need urgent balloon septostomy to enlarge ASD
Interrupted aortic arch	Rapid breathing, sleepiness, tachycardia, feeding difficulties	Within the first few days of birth	• Open heart surgery ASAP
TOF	Cyanosis, difficulty breathing with feeding, clubbing, poor weight gain, problems eating, prolonged crying, developmental delay, irritability, heart murmur	Often diagnosed prenatally or within hours to days of birth	• Surgery to shunt more blood to the lungs and temporarily repair • Complete repair of 2 of the 4 abnormalities

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Table 3
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	Presentation	Timing	Intervention
Pulmonary atresia	• Pulmonary blood flow may be supplied by patent ductos arteriosus or MAPCAs and have more mild symptoms	Variable • If severe will be cyanotic in the first hours or days of life as the DA starts to close and can lead to circulatory collapse • If mild degree of outflow obstruction may have heart failure symptoms initially from left to right shunt but with hypertrophy of the right ventricle infundibulum as the patient grows, cyanosis will occur in the first few months of life	• Depends on the anatomy but some can be treated with cardiac catheterization and valvuloplasty others require open heart surgery for repair with a valve or shunt from the aorta to the pulmonary artery • Multiple surgeries are often needed
Tricuspid atresia	• Depends on the degree of pulmonary obstruction, moderate will have decreased pulmonary blood flow and cyanosis • If large VSD and minimal RVOT obstruction, may present with signs of pulmonary over circulation and heart failure	Moderate degree of PS symptomatic in the first days to weeks of life	Severely cyanotic neonates require PGE until aortopulmonary shunt can be placed (preferred anastomosis or Blalock-Taussig procedure) • Bidirectional glen shunt between 2 and 6 mo of age decreases the chance of left ventricle dysfunction in life
Ebstein anomaly of tricuspid valve	Cyanosis, massive cardiomegaly, and long holosystolic murmur	Newborn	Several surgical options depending on severity of hypoxia including aortopulmonary shunt, atrial septectomy, surgical path of the tricuspid, "one and a half ventricle repair"

Blue, right heart obstructive lesions; Green, left heart obstructive lesions; White, mixing lesions.

Abbreviations: ASD, atrial septal defect; D-TGA, dextro-transposition of the great arteries; PGE_1, prostaglandin E1; RVOT, right ventricular outflow tract; TAPVR, total anomalous pulmonary venous return; TOF, tetralogy of fallot; VSD, ventricular septal defect.

Data source [Bernstein, Daniel. Chapter 461: General Principles of Treatment of Congenital Heart Disease. In: Nelson Essentials of Pediatrics, 21st Edition. Philadelphia, PA: Elsevier; 2020. P 2428-2436.e1].

CYANOTIC, LEFT HEART OBSTRUCTIVE LESIONS

Hypoplastic Left Heart Syndrome

In HLHS, the left ventricle, mitral valve, and aorta do not develop normally, and infants present with cyanosis, respiratory distress, and signs of low cardiac output. Cyanosis is a result of mixing oxygenated and deoxygenated blood. Atrial septal defect (ASD) is required to support blood flow to the left side of the heart and balloon atrial septostomy may be needed urgently if a restrictive ASD is present. PGE is part of the treatment because respiratory distress and low cardiac output often result from ductal constriction or restrictive ASDs. PGEs will be administered until a surgical procedure called the Norwood procedure can be completed.

Interrupted aortic arch

This type of cyanotic lesion is more likely than most heart problems to be missed with POS and presents with shock in the first week of life. The treatment recommended is PGE with consideration of fluorescence in situ hybridization (FISH) for the evaluation of DiGeorge syndrome.

CYANOTIC, RIGHT HEART OBSTRUCTIVE LESIONS

Many cyanotic lesions are due to the right ventricular outflow tract (RVOT) obstruction. Not all problems with right heart outflow result in cyanosis. The degree of cyanosis depends on the severity of pulmonary obstruction.

Tetralogy of Fallot

TOF is in the conotruncal family of heart lesions where the primary problem is anterior deviation of the muscular septum that separates the aortic and pulmonary outflows. This anomaly results in 4 problems with varying degrees of severity: (1) obstruction to the RVOT (pulmonary stenosis [PS] and in extreme cases, atresia), (2) ventricular septal defect (VSD), (3) dextroposition of aorta so that it overrides the ventricular septum, and (4) right ventricular hypertrophy. Cyanosis may be progressive, and the patient is at risk of "tet" spells. Patients often present with a loud systolic murmur heard best in the upper sternal boarder. PGE are sometimes used to maintain the patency of the ductus in neonates with TOF with pulmonary atresia as part of early treatment. FISH testing (22q11) should be considered due to the association with DiGeorge syndrome or velocardiofacial syndrome (also known as CATCH 22: cardiac defects, abnormal facies, thymic hypoplasia, cleft palate hypocalcemia). Infants are at risk of developing progressive sub-PS, which is the cause of the "tet spells" in the first few months of life. During a tet spell, infants should be soothed, given oxygen and positioned with knees to chest to shunt blood toward the lungs. When hyperchaotic episodes are observed, transfer to a pediatric cardiologic surgeon for prompt repair is the important next step. Growth, development, and puberty may also be delayed in patients who have not undergone surgery and are chronically hypoxic (Sa O_2 <70%).

Pulmonary Atresia

Pulmonary atresia is complete obstruction of the RVOT. The pulmonary blood flow is variable and could be dependent on PDA or have multiple major aortopulmonary collateral arteries (MAPCAs) originating from the ascending and/or descending aorta and supplying various lung segments. Pulmonary atresia, also known as critical pulmonic stenosis, can result in right-sided heart failure (hepatomegaly, peripheral edema) and cyanosis from right to left shunting across a patent foramen ovale.

Tricuspid Atresia

In tricuspid atresia, the valve between the right atrium and the right ventricle fails to form, resulting in a variable degree of cyanosis. An infant with tricuspid atresia can present with shock if there is a restrictive VSD.

Ebstein Anomaly of Tricuspid Valve

Ebstein anomaly is due to downward displacement into the right ventricle of an abnormal tricuspid valve. The right atrium is often enlarged due to tricuspid regurgitation. The output from the right heart decreases due to a combination of poorly functioning ventricle, tricuspid regurgitation, and RVOT obstruction from the anterior leaflet of the tricuspid valve being enlarged. Blood shunts from the right to left atrium through the foramen ovale leading to cyanosis. In severe cases, the force to open the right ventricle may not be strong enough, which results in functional pulmonary atresia. The left atrium blood mixes with blood returning from the pulmonary veins. When the PDA closes, severe cyanosis may develop. The degree of symptoms varies greatly depending on the severity of the RVOT.

CYANOTIC, MIXING LESIONS

Truncus Arteriosus

Truncus arteriosa\us results when the aorta and pulmonary arteries do not completely separate leading to mixing of oxygenated and unoxygenated blood. Some children will develop cyanosis with truncus arteriosus due to obligate mixing of oxygen.

Transposition

Transposition of the great arteries occurs when the aorta comes out of the right ventricle and the pulmonary artery from the left ventricle and both systemic and pulmonary circulation operate independently without crossing each other. Treatment urgency of this lesion depends on whether the ASD is restrictive, which requires urgent balloon atrial septostomy to enlarge the ASD or if adequate mixing is occurring due to ASD.

Total Anomalous Pulmonary Venous Return

With TAPVR, the connections of all 4 pulmonary veins are abnormal. In the first few hours of life, there is an increased perfusion of the lungs resulting in cyanosis, respiratory distress, and signs of low cardiac output. If the pulmonary veins are not obstructed, infants may have minimal symptoms. This is one of the few surgical emergencies in pediatric cardiology, which requires urgent surgical treatment. It can worsen with PGE.

Acyanotic congenital heart disease

There are several types of noncyanotic CHD as described in **Table 4**. There are 2 main subtypes of noncyanotic CHD, differentiated by the effect on the heart. One subset results in an *increased volume* of blood in the heart due to the effect of the left-to-right shunt. These include the ASD, atrioventricular septal defects (VSD, atrioventricular [AV] canal), and patent ductus arteriosus (PDA) lesions. The other subset results in an *increased pressure* load in the heart. These are the "ductal-dependent" lesions and are most commonly the AS, CoA, and PS.

NON-CYANOTIC, LEFT-TO-RIGHT CONGENITAL HEART DISEASE

The lesions resulting in an increased volume of blood in the heart have one thing in common—they shunt blood from the higher pressure left side of the heart to the right side of the heart through a patent opening. This shunting of blood results in higher

Table 4
Physical findings associated with noncyanotic heart disease (for isolated lesions)

	Murmur Type	Location	Radiation	Thrill	Pulse Findings	Other Findings
ASD	May be none, systolic ejection	Second intercostal space	Not usually	Not usually	No changes	Fixed split S2
PDA	Initially systolic, becoming continuous systolic/diastolic	Under left clavicle	Back	With large left-to-right shunt	May have bounding pulses	Ejection clicks May have cyanosis/ clubbing of the LE only
VSD	Harsh, blowing holosystolic May have diastolic rumble	Left sternal border	None	Maybe at left lower sternal border	No changes	Murmur may appear in the first week of life May have S3
AV Canal	Variable and dependent on the type of AV canal defect					
AS	Systolic ejection, harsh, crescendo-decrescendo May have murmur of diastolic aortic insufficiency	Right upper sternal border Sternal notch	Neck	Possibly at sternal notch or right base	May be decreased	May have hyperactive precordium May have ejection click
CoA	May be none. Systolic harsh sounding	Back, between scapula	Not usually	Not usually	Deceased pulses—all 4 extremities or decreased/delayed in LE	Pulse ox in R UE higher than LE
PS	Systolic ejection	Left upper sternal border	Maybe to the back or axillae	Possibly	Routine	Click after S1 Split S2

Data source [Marcdante KJ, Kliegman RM, Schuh AM. Chapter 143: Acyanotic Congenital Heart Disease. In: Nelson Essentials of Pediatrics, 9th Edition. Philadelphia, PA: Elsevier; 2023. P 564-8.]

blood volumes in the heart as left-sided, oxygenated blood intermixes with right-sided, deoxygenated blood, resulting in increased volume of blood to the right side of the cardiovascular system. If a clinically significant lesion is present, over time this increase in right-sided volume can decrease pulmonary compliance, resulting in symptoms of overload including tachypnea, tachycardia, retractions, flaring, wheezing, and failure to thrive. Some lesions over time will result in increased pulmonary resistance, resulting in a reversal of the shunt to right-to-left. Some infants born with these lesions will initially have no murmur but one can develop over time (after 6 hours of life) as pulmonary resistance drops after birth, increasing the pressure gradient.

Atrial Septal Defects

An ASD occurs when the atrial septal tissue fails to fully develop in utero, resulting in a "hole in the heart" between the left and right atria. There are 4 subtypes of ASD. The ASD may be an isolated finding or associated with other cardiac abnormalities. When associated with other cardiac abnormalities, the other findings are usually more clinically relevant. The secundum ASD is the most common subtype and are also the type most likely to spontaneously resolve. ASD is different from patent foramen ovale (PFO), where the FO does not fully close after the newborn period. The FO is part of the fetal circulatory system allowing oxygenated blood from the right atrium to pass into the left atrium and then out through the left side of the heart the peripheral circulation. After birth, the FO closes, and when the closure is incomplete, it results in a PFO that is often clinically silent. Presentation of ASD may vary. The murmur of an ASD is typically (mid)systolic with a crescendo-decrescendo quality, most found at the right upper sternal border. Small secundum ASD lesions can close spontaneously; however, larger ASD are most likely to persist, resulting in increased right-sided cardiac pressures. These are typically well tolerated until around 40 years of age, when cardiac symptoms develop. Complications include volume overload, pulmonary hypertension, exercise intolerance, heart failure, and arrhythmias. ASD that does not close spontaneously is treated via surgical intervention, either via open-heart surgery or via a cardiac catheterization.

Patent Ductus Arteriosus

The ductus arteriosus (DA) is a vascular connection found between the pulmonary artery and the aorta, distal to the left subclavian artery. After birth, the DA starts to constrict and is functionally closed within 10 to 15 hours of life, with complete closure by age 2 to 3 weeks. The DA closure is mediated by a decrease in circulating PGE. In some infants, the DA remains patent. This results in the continued shunting of blood but due to the decreased pulmonary resistance, the blood flow is from the aorta to the pulmonary artery, creating a left-to-right-sided shunt. The clinical manifestations are determined by the degree of shunting. Small PDA may result only in a continuous systolic murmur. Medium-sized PDA may present with symptoms of exercise intolerance, a continuous systolic/diastolic murmur, wide pulse pressure, and left ventricular overload. Large PDA can result in left-sided volume overload leading to an increased pulmonary resistance and reversal of the shunt to right to left, and these patients eventually become cyanotic (infants can present with heart failure early in life). Other physical findings include clubbing of the lower extremities, a dynamic apical impulse, a palpable thrill, and bounding pulses due to wide pulse pressures. If left untreated, patients with PDA are at risk for heart failure, pulmonary hypertension, and rarely, infective endocarditis. The treatment of PDA may be medical with use of prostaglandin inhibitors or surgical with either ligation or transcatheter closure.

Ventricular Septal Defects

A VSD occurs when the ventricular foramen does not close resulting in a left-to-right shunt through a patency between the ventricular chambers. The size of the defect determines the degree of the shunt and the extent of the increased volume to the right side of the heart. Newborns typically do not have a murmur at birth but one manifests within the first week or so of life. The murmur is typically holosystolic, and newborns with large VSD may present with symptoms of heart failure within the first month of life. Complications of VSD include pulmonary hypertension, infective endocarditis, aortic regurgitation, arrhythmias (including atrial fibrillation), heart failure, atrial shunting, sub-AS, and RVOT obstruction. Some small-to-moderate-sized VSD close spontaneously. Patients with small VSD and no symptoms may be watched clinically. For those that are symptomatic, medical therapy for heart failure is indicated, and some require surgical closure.

AV Canal Lesions

AV canal defects (also called AV septal defects, endocardial cushion defects, or persistent AV ostium) are a group of congenital heart conditions that involve the AV septum (atrial only or both atrial and ventricular) and the AV valves (mitral and tricuspid). There is a strong correlation between AV canal defects and Down syndrome. There are 4 different subtypes—the complete and intermediate subtype have both ASD and VSD with different valvular pathologic condition but have similar physiology. Transitional AV canal defects have an insignificant, small VSD so physiologically behave similarly to the partial defect (ASD only). Complete AV canal defects result in significant left-to-right shunting, resulting in increased pulmonary blood flow and heart failure symptoms by 1 year of age. Pulmonary hypertension may result. Partial AV canal defects are associated with valvular issues, most commonly mitral regurgitation. These lesions are present in a similar fashion to ASDs. These patients may have minimal symptoms until adulthood. The treatment of AV canal defects is surgical, with attention to both the septal defects and any associated valvular issues.

NONCYANOTIC, DUCTAL-DEPENDENT CONGENITAL HEART DISEASE

Noncyanotic, ductal-dependent lesions have an obstruction to the normal cardiac blood flow. These lesions may be present at birth, shortly after birth or later in life. They can result in left-sided or right-sided heart failure, or other symptoms, depending on the lesion.

Aortic Stenosis

The most common form of AS is at the valvular level with the aortic valve having 2 leaflets instead of 3. These lesions are generally detected on prenatal ultrasound. Critical AS presents when the closure of the DA occurs, leading to inadequate cardiac output and the development of heart failure. A murmur is often not appreciated because the cardiac output is too low. Older children with AS are usually asymptomatic and may have an ejection click with a systolic ejection murmur. The murmur is louder with more significant stenosis, has a crescendo-decrescendo, harsh quality, and there may be an aortic regurgitation diastolic component. Over time, a thrill may be appreciated. The AS tends to progress over time, and there is an increased risk for sudden death, infective endocarditis, and the development of aortic regurgitation. Treatment of AS ranges from antenatal treatments to valvuloplasty, valvotomy, and valvular replacement.

Coarctation of the Aorta

When a patient has a narrowing of the descending aorta, typically just proximal to the insertion of the DA, they have CoA. After birth, when the DA and foramen ovale begin to close, the CoA causes increased left-sided pressures. Clinically this can appear as "critical coarctation" where the infant presents in heart failure as the DA closes. Their lower extremity pulses may be diminished or absent and they may have hepatomegaly. Pulse oximetry will be higher in the upper extremities compared with the lower. Infants with critical coarctation, once identified, should receive intravenous prostaglandin to maintain their PDA, as well as heart failure treatment before definitive repair. Infants with less-severe CoA may have a cardiac murmur (systolic murmur heard in the back, harsh in quality) and decreased femoral pulses. Heart failure does not typically occur outside of the newborn period. Treatment of CoA may be transcatheter or surgical in nature, and the modality depends on the age of the patient, the degree of the CoA, and associated cardiac conditions.

Pulmonary Stenosis

As opposed to AS, which is a left ventricular outflow lesion, PS is a right ventricular outflow lesion. Valvular PS is the most common form of PS. PS causes outflow obstruction to the pulmonary arteries, resulting in right ventricular hypertrophy, the degree of which is directly related to the degree of stenosis. Most cases of PS are not discovered until after birth. Infants with severe PS will present soon after birth when the obstruction causes right-sided outflow obstruction, with a right to left shunt when the DA starts to close, resulting in increased right-sided pressures, and the newborn may go into heart failure and seem cyanotic. Less-severe obstruction may be picked up by screening pulse oximetry or with an audible systolic ejection murmur. Most infants and children are asymptomatic. Over time, patients may develop shortness of breath and fatigue due to increased right-sided cardiac outflow obstruction. The treatment of PS depends on the severity of the lesion. Newborns with critical PS require PGE to keep the DA patent, and once stable, they require definitive treatment with balloon valvuloplasty dilatation or surgical intervention. Mild PS is usually watched clinically, whereas more significant PS usually warrants procedural intervention.

DISCUSSION

Patients with CHD have complex health needs and require primary care providers to promote care coordination with specialty services and assist in smooth transitions of care.[23] Children with CHD often have prolonged hospitalizations and account for 15.1% of all hospital costs for pediatric patients.[24] Children living with CHD also require more educational services.[24] A primary care provider's role includes support not only for the child but also for the family.[9] Family support includes helping families navigate the stresses of having a child with a chronic health condition as well as providing resources for families to recognize and manage potential issues unique to CHD including caregiver training in cardiopulmonary resuscitation and automated external defibrillator use.[9]

Children with CHD can have rapid decompensation during respiratory or gastrointestinal infections from changes in intravascular volume.[9] This risk is especially true for more severe CHD. Children with DiGeorge syndrome and associated CHD are not recommended to receive live vaccines. Immunoprophylaxis against respiratory syncytial virus can be considered for children less than 12 months who have hemodynamically significant CHD.[5]

Growth and Nutrition

Children with CHD have unique nutritional considerations because they may have higher caloric needs (120–150 kcal/kg/d) for adequate growth.[23] To achieve this growth, some infants may require fortification of formula or expressed breast milk to 30 kcal/ounce. Growth issues for children with CHD can be multifactorial but may include poor caloric intake, high metabolic demands, and other genetic or extracardiac abnormalities.[25]

Endocarditis Prevention

Prevention of endocarditis for children and adults with CHD has been updated to have fewer indications for antibiotic prophylaxis. Antibiotic prophylaxis is indicated for dental procedures involving manipulation of gingival tissue, periapical region of the teeth, or perforation of the oral mucosa for the following patients:[23,26]

- Prosthetic cardiac valve or prosthetic material used for cardiac valve repair
- Cardiac transplantation recipients who develop cardiac valvulopathy
- CHD specifically
  - Unrepaired cyanotic CHD including palliative shunts and conduits
  - Complete repaired of CHD with prosthetic material or device during first 6 months only
  - Repaired CHD with residual effects

Development and Academic Performance

Children with CHD have ongoing developmental needs and are 50% more likely to receive special education services while in school.[2] It is not clear why there are notable developmental delays in children with CHD. Contributors may include prolonged hospitalizations as infants, restrictions that follow surgery, the association of CHD with genetic syndromes, or specific CHD diagnoses with worse motor impairment.[3,27] One-third of children with CHD have delayed motor skills, and there is a higher prevalence of lower intellectual abilities and behavioral difficulties that extends into adolescence and adulthood.[3] Updated guidelines from the American Heart Association recommend implementing standardized screening and evaluation of motor skills for children and adolescents living with CHD.[28]

Participation in Sports

For children without hemodynamically significant CHD lesions, physical activity and sports participation are recommended. Participation in activity is often recommended for these patients to avoid being sedentary and reduce the risk of obesity and hypertension. Conditions at highest risk during exercise are below but require careful consideration by the primary care provider and the cardiologist.[23]

- Severe ventricular outflow obstruction
- Hypertrophic cardiomyopathy
- Congestive heart failure
- Coronary insufficiency
- Pulmonary hypertension
- Severe untreated systemic hypertension
- Marfan syndrome and aortic dilation
- Exercise-induced arrhythmia
- Long QT syndrome

SUMMARY

More people are living with CHD because many children now survive to adulthood with advances in medical and surgical treatments. Patients with CHD have ongoing complex health-care needs in the various life stages of infancy, childhood, adolescence, and adulthood. Primary care providers should collaborate with pediatric specialists to provide ongoing care for people living with CHD and to create smooth transitions of care.

CLINICS CARE POINTS

- Children with CHD may require consultation with a geneticist if there are concerns for a genetic abnormality.
- Fetal echocardiogram can be used to identify some CHD lesions in the prenatal period but does not exclude all CHD.
- Pulse oximetry is a useful tool to screen newborns for CHD but normal pulse oximetry does not exclude all CHD.
- Children with CHD aged younger than 12 months may be eligible for immunoprophylaxis against respiratory syncytial virus.
- Children with CHD require careful monitoring for appropriate growth, particularly in infancy.
- Standardized screening and evaluation for motor development delays should occur for all children with CHD.
- Children with CHD require collaboration between the primary care provider, pediatric cardiologist, and surgeon (if involved).
- All providers involved in the care of children with CHD should assist in creating smooth transitions of care.

DISCLOSURE

None.

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