Consultation with the Specialist

Evaluation of Heart Murmurs

J. Peter Harris, MD*

Introduction

As many as 90% of children will have an audible heart murmur at some point in time. However, the prevalence of nontrivial heart disease in this age group is less than 4 per 1000. The practitioner's task is to distinguish the ubiquitous innocent or normal murmur from the murmur indicating heart disease. Normal murmurs include vibratory and pulmonary flow murmurs, venous hums, carotid bruits, and the murmur of physiologic branch pulmonary artery stenosis. Mislabeling of a normal murmur as pathologic may have adverse psychological effects on the family and the child, including unwarranted exercise restrictions and problems later with insurability and employment. Alternatively, failure to identify a pathologic murmur may delay appropriate intervention, especially in early infancy. Recent well-publicized sudden cardiac deaths of professional athletes have augmented the anxiety caused by the specter of heart disease in the young.

Because less than 5% of heart murmurs in children denote cardiac pathology and because each practitioner is likely to see no more than five patients per year who have important heart disease, distinguishing normal from organic murmurs is not always simple. Nevertheless, given the high prevalence of murmurs in the young and because nearly 50% of the patients who have murmurs referred to pediatric cardiologists do have underlying heart disease of variable severity, most clinicians can recognize normal murmurs. The remaining referred patients have murmurs ultimately judged to be normal but that may have been atypical or not readily identified in a busy pediatric practice.

Associate Professor of Pediatric Cardiology, University of Rochester School of Medicine and Dentistry, Rochester, NY.

History

The first step in the evaluation is a careful history. Symptoms of exercise intolerance, feeding difficulties, dyspnea, cyanosis, or syncope should alert the clinician to the potential of cardiac dysfunction. Other symptoms that at least raise the possibility of organic heart disease include failure to thrive, diffuse diaphoresis, unexplained persistent irritability or lethargy, and atypical chest pain. Although symptoms are helpful if present, the majority of young patients who have heart murmurs are asymptomatic. However, most critical cardiac malformations in early infancy are characterized by persistent peaceful tachypnea (a respiratory rate greater than 60 breaths/min in a young infant), a consequence of either pulmonary venous congestion or hypoxemia. An inquiry about the breathing pattern of a young infant, especially compared with older siblings at a similar age, is worthwhile. The patient's gestational course should be reviewed, particularly in regard to exposure to potential teratogens and maternal illnesses. For instance, fetal exposure to lithium may cause Ebstein anomaly of the tricuspid valve. Ventricular as well as atrial septal defects are well-known components of the fetal alcohol syndrome. In addition to a transient hypertrophic cardiomyopathy, infants of diabetic mothers may present infrequently having a tetralogy of Fallot, truncus arteriosus, or double outlet right ventricle. Both overt and occult maternal collagen vascular disease may lead to the development of fetal complete heart block.

A thorough family history also should be obtained. The preoccurrence of a congenital cardiovascular malformation increases the risk of a cardiac defect. Early epidemiologic studies suggested a multifactorial basis of inheritance for nearly 90% of cardiac anomalies, with a recurrence risk of 1% to 4%. More complete prospective investigations using echocardiography and classification by pathogenetic mechanisms are now revealing substantially higher recurrence rates, especially among families in which left heart obstructive lesions occur. Therefore, the concept of multifactorial inheritance is becoming less attractive. Rather, many cardiovascular malformations may be the result of single gene defects. For example, a deletion in chromosome 22, subband q11, has been found in patients who have DiGeorge syndrome (type B interrupted aortic arch, truncus arteriosus) and the velo-cardiofacial syndrome (tetralogy of Fallot with pulmonary atresia). If many cardiac malformations do have a genetic basis, survival into childbearing age of women who have had defects repaired will increase the population having congenital heart malformations.

Physical Examination

Recognition or suspicion of a syndrome increases the likelihood of heart disease (Table 1). Similarly, because 25% of children who have heart disease have extracardiac anomalies, the presence of noncardiac malformations should increase the level of suspicion for organic heart disease. Among infants who had major gastrointestinal malformations (diaphragmatic hernia, tracheoesophageal fistula and esophageal atresia, omphalocele, and imperforate anus) and were referred to a tertiary medical center, congenital cardiac defects were found in 15% to 25%. The most common cardiac malformations

were ventricular septal defect and tetralogy of Fallot. Because these reviews were not population-based and, therefore, subject to ascertainment bias, the exact incidence of congenital cardiac defects in patients who have extracardiac anomalies is unknown. Nevertheless, clinicians need to be aware of the risk of cardiovascular problems in patients who have noncardiac malformations as well as associations with defined syndromes.

The infant or child who has cyanosis not attributed to respiratory dysfunction should be referred for cardiovascular evaluation, regardless of the presence or absence of a murmur. The same rule applies to patients who have an unexplained abnormal rate or pattern of breathing, a persistently hyperdynamic precordium, precordial bulging, or asymmetric pulses. Signs of congestive heart failure—an inappropriate tachycardia, tachypnea, firm and often tender hepatomegaly, and an abnormal pulse volume—also should prompt a referral to a pediatric cardiologist. The task of recognizing serious heart disease is more arduous if such signs are absent or subtle. Auscultation then becomes of paramount importance, although the situation is complicated by the prevalence of normal murmurs and, not infrequently, the absence or subtle nature of murmurs in newborns and young infants who have serious cardiac disease.

In the absence of associated cardiovascular abnormalities, recognized syndromes, and extracardiac malformations, ausculatory criteria signifying cardiac disease include: loud, pansystolic, late systolic, diastolic, or continuous murmurs; an abnormally loud or single second heart sound; a fourth heart sound or S_4 gallop; and

Table 1. Selected Syndromes and Associated Cardiac Malformations

SYNDROME	INCIDENCE OF CARDIAC MALFORMATIONS (%)	CARDIAC MALFORMATIONS
Down	50	AVSD, VSD, ASD, PDA, TOF
Trisomy 18	99	VSD, PDA, DORV, BPV
Trisomy 13	90	VSD, ASD, PDA
Turner	40	Coarc, AVS, HLH
Noonan	50	PVS, HCM
William	90	SVAS, SVPS, RAS
Marfan	60 to 80	MVP, AoRD, AI
DiGeorge	90	IAA(B), TA
VACTERL	80	VSD, ASD, PDA, TOF

AVSD = atrioventricular septal or canal defect, VSD = ventricular septal defect, ASD = atrial septal defect, PDA = patent ductus arteriosus, TOF = tetralogy of Fallot, DORV = double outlet right ventricle, BPV = bicuspid pulmonary valve, coarc = coarctation, AVS = aortic valve stenosis, HLH = hypoplastic left heart, PVS = pulmonary valve stenosis, HCM = hypertrophic cardiomyopathy, SVAS = supravalvular aortic stenosis, SVPS = supravalvular pulmonary stenosis, RAS = renal artery stenosis, MVP = mitral valve prolapse, AoRD = aortic root dilatation, AI = aortic insufficiency, IAA (B) = interrupted aortic arch type B, TA = truncus arteriosus.

LESION	SHAPE	TIMING	LOCATION	OTHER FINDINGS
Ventricular septal defect (VSD)	Plateau	Holosystolic	LLSB	Apical mid-diastolic murmur with large shunt
Mitral regurgitation	Plateau	Holosystolic	Apex	Higher pitched than VSD murmur
Atrial septal defect	Ejection	Systolic	ULSB	Persistent S_2 split
Patent ductus arteriosus	Diamond	Continuous	ULSB	Bounding pulses
Aortic valve stenosis	Ejection	Systolic	URSB	Ejection click
Subvalvular aortic stenosis	Ejection	Systolic	ML-URSB	No ejection click
Hypertrophic cardiomyopathy	Ejection	Systolic	LLSB-apex	Laterally displaced PMI
Coarctation	Ejection	Systolic	ULSB-Left back	Pulse disparity
Pulmonary valve stenosis	Ejection	Systolic	ULSB	Ejection click; wide S_2 split
Tetralogy of Fallot	Ejection	Systolic	MLSB	Cyanosis

LLSB = lower left sternal border, ULSB = upper left sternal border, URSB = upper right sternal border, MLSB = mid-left sternal border, S_2 = second heart sound, PMI = point of maximal impulse.

ejection or midsystolic clicks. A physiologic third heart sound may be audible in healthy children in the supine position but usually disappears with assumption of an upright posture. An infant or child who has any of these findings should be referred for evaluation.

Although relatively characteristic murmurs do occur with cardiac malformations, as outlined in Table 2, physiologic variations alter the auscultatory findings. Consider the infant who has a ventricular septal defect (VSD). The typical murmur heard in the presence of a medium-size or restrictive VSD is a harsh pansystolic murmur of even amplitude that is audible at the lower left sternal border. Murmurs are caused by turbulence that usually is generated by a difference in pressure. Hence, in the presence of a restrictive VSD, a holosystolic pressure gradient from the left ventricle (eg, 90 mm Hg) to the right ventricle (eg, 45 mm Hg) will produce a holosystolic murmur. However, if the defect is large or nonrestrictive, ventricular pressures are equal. Because no pressure gradient exists, no murmur is generated. This clinical picture often is responsible for the delay in detection of large ventricular defects after birth. However, the second heart sound will be difficult to split and will be accentuated due to a loud pulmonary component. Attention to the second heart sound and the finding of a hyperdynamic precordium may allow earlier detection. Alternatively, in the infant who has a tiny or pinhole-size ventricular defect, the murmur will be quite soft and short, ending well before the second heart sound, which is split and of normal intensity.

A similar quandary occurs with the patient who has a patent ductus arteriosus (PDA). The typical murmur of a restrictive PDA is continuous, louder in systole, and located at the upper left sternal border. However, the patent ductus in small preterm infants usually is large or nonrestrictive. A torrential left-to-right shunt may develop in the absence of a murmur or with only a soft systolic component, but bounding pulses and a hyperdynamic precordium are clues to the presence of a PDA. If a continuous murmur is noted, the clinician needs to rule out an obligatory or necessary ductus, as seen in neonates who have pulmonary atresia or a coarctation of the aorta. Is the patient cyanotic? Is there a pulse discrepancy? Either of these findings dictates an urgent referral.

In contrast to holosystolic plateau murmurs seen with a VSD or mitral regurgitation, ejection (diamond-shaped, crescendo-decrescendo) murmurs are caused by ventricular outflow obstruction. In infants who have fast heart rates, this differentiation may be difficult. Ejection murmurs begin after the first heart sound in contrast to pansystolic murmurs, which begin at or are coincident with the first heart sound. The most common varieties, aortic and pulmonary valvular stenosis, usually are associated with an early systolic, high-pitched, ejection click due to sudden doming of the valves. Such clicks occur at the onset of the murmur and are best appreciated between the lower left sternal border and the cardiac apex in aortic stenosis and at the upper left sternal border in pulmonary valve stenosis. With increasing severity of obstruction, ejection murmurs become louder. However, the converse occurs in patients who have subvalvular pulmonary stenosis in tetralogy of Fallot. As the obstruction worsens in a tetralogy, the murmur becomes softer and shorter, but a concurrent increase in cyanosis usually is seen.

It is beyond the scope of this review to discuss all of the organic heart murmurs encountered in young patients. The clinician does need to be aware of the vicissitudes of pathologic murmurs, but the practitioner's primary task is to differentiate normal from pathologic murmurs.

A normal heart murmur can be diagnosed if: the patient is asymptomatic; there is no evidence of associated cardiac abnormalities, extracardiac congenital malformations, or syndromes; and there are characteristic auscultatory features of an innocent murmur. For example, a healthy-appearing, acyanotic, nondysmorphic 18-month-old infant who has a grade 2, low-pitched, musical, midsystolic ejection murmur at the lower left sternal border and an otherwise normal cardiovascular evaluation has a typical vibratory murmur. Similarly, a left-sided venous hum can be distinguished from a continuous murmur of a PDA by disappearance of the hum with the assumption of a supine posture. Venous hums frequently are louder in diastole, are of medium pitch, and may be abolished by compressing the jugular vein. A pulmonary flow murmur with a physiologically split second heart sound but no other cardiovascular findings in a healthy adolescent should be labeled as normal. The characteristics of normal murmurs are summarized in Table 3. It is important to remember that normal murmurs may coexist in the same patient. Careful auscultation, using the techniques of "inching" and "dissection" (concentrating on one segment of the cardiac cycle at a time), usually will clarify the clinical picture. If an innocent murmur is detected, the parents should be informed that their child's heart is indeed normal, no restrictions or special precautions are necessary, and no follow-up is required.

Between the clearly pathologic and obviously normal ends of the spectrum lies a gray area, the not-clearly innocent murmur, which constitutes the major source of referrals to the pediatric cardiologist. When confronted with this situation, the practitioner can elect one of several strategies, dictated in large part by his or her training, experience, and level of comfort as well as by the parent's degree of anxiety. If the index of suspicion for heart disease is low and the examination is thwarted by anxious behavior, with its attendant tachycardia, auscultation may be repeated after the child calms or at a later date. For example, tachycardia tends to obscure the characteristic low-pitched, mid-systolic, musical, ejection quality of a vibratory murmur. A repeat examination when the child is quiet and supine, both of which promote cardiac slowing, frequently will allow the classic features of a vibratory or Still murmur to become evident. Similarly, the transient murmur of physiologic branch pulmonary artery stenosis (PPS) may not be identified readily in a young infant whose state of activity varies throughout the examination. The characteristic finding of PPS—a relatively high-pitched, ejectile murmur of equal but soft intensity throughout the chest—usually is evident in the

TYPE	SHAPE	TIMING	PITCH	LOCATION	OTHER FINDINGS
Vibratory	Ejection	Midsystolic	Low	LLSB-apex	Intensity ≤ grade II
Venous hum	Diamond	Continuous	Medium	Subclavicular	Disappears in supine position
Carotid bruit	Ejection	Systolic	Medium to low	Base of neck	Decreased by carotid compression
Pulmonary flow	Flow	Systolic	Medium	ULSB	Normal S_2 split
Physiologic branch and pulmonary artery stenosis	Ejection	Systolic	Medium	Entire chest	Disappears by 4 to 6 months of age

LLSB = lower left sternal border, ULSB = upper left sternal border, S_2 = second heart sound.

infant who remains in the same quiet physiologic state during auscultation. The strategy of ordering an echocardiogram followed by referral to a cardiologist if abnormalities are found should be discouraged because of its expense and the possibility that the echocardiogram may not be performed or interpreted by a trained pediatric cardiologist. The majority of normal murmurs are identified readily as such through auscultation by the cardiologist without the need for costly additional testing. Finally, if heart disease is suggested or parental anxiety is extremely high, referral to a pediatric cardiologist is appropriate.

The Newborn Infant: A Special Situation

Recognition of heart disease in a neonate often is difficult. To begin with, depending on the frequency of auscultation, up to 60% of healthy term newborn infants have normal murmurs. Additionally, one third of neonates who have serious heart malformations may not have a detectable heart murmur during the first 2 weeks of life. Thus, the presence of a murmur is not a specific indicator of cardiac pathology, and the absence of a murmur does not guarantee cardiac well-being. In fact, up to 30% of newborn infants subsequently judged to have heart disease are discharged from the newborn nursery as ostensibly healthy. Late referral of a young infant who has critical heart disease frequently engenders practitioner consternation and parental dissatisfaction. Therefore, the clinician should pay attention to other, frequently subtle cardiovascular signs in the neonate. For example, regardless of whether a murmur is present, persistent peaceful tachypnea, either by report from the family or by clinical observation, should not be dismissed; 90% of infants who have serious cardiac disease referred after 2 weeks of age have had persistent tachypnea with onset soon after birth. In this current era of early neonatal discharge, a discussion of persistent tachypnea may be a worthwhile component of anticipatory guidance. In addition, the detection of a persistently hyperdynamic precordium should alert the practitioner to the possibility of organic heart disease.

Auscultation of the second heart sound, a previously ignored physical finding, also may help. In the overwhelming majority of healthy and quiet term neonates, the second heart sound is split audibly by 12 hours of age and often earlier. In general, a split second heart sound signifies the presence of two semilunar or outflow tract valves as well as an appropriate reduction in postnatal pulmonary vascular resistance. A single second heart sound in a quiet neonate indicates: 1) the absence of one semilunar valve, as in aortic or pulmonary atresia; 2) an abnormal position of the great vessels, as in transposition of the great arteries or tetralogy of Fallot; or 3) pulmonary hypertension, as seen with a large ventricular defect or persistent pulmonary hypertension of the newborn. Auscultation of the second heart sound is of little value in the infant who has tachycardia, but the sleeping heart rate of term neonates is approximately 100 beats/min, a rate at which most practitioners can recognize splitting of the second heart sound. The vast majority of neonates who have critical heart disease have an audibly single and often accentuated second heart sound. Thus, auscultation of the second heart sound in sleeping or quiet neonates is an additional screening modality that may enhance early detection of serious cardiac malformations.

SUGGESTED READINGS

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