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Short Communication

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Doctor, is there Something Wrong with my Baby's Heart? – A Clinical Approach

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Abstract:

Congenital heart disease (CHD) is the most common birth defect. Abnormalities range from trivial to mild to potentially fatal lesions. The diagnosis of CHD is aided by prenatal ultrasound screening, usually conducted between the first and second trimesters with good cardiac imaging obtainable at 20-22 weeks gestation. Although such screening is invaluable it still has limitations and tends to be operator dependent. It may however detect most of the serious CHD, prewarning clinicians and permitting in utero transfer for those requiring neonatal care. On assessing the newborn, a cardiac abnormality is suggested by the presence of a murmur, tachypnoea, cyanosis, and/or differential pulses. It is essential to distinguish between cardiac and non-cardiac causes of tachypnoea and cyanosis. Pulse oximetry screening has further improved the detection of potentially serious CHD in addition to recognizing neonates with mild oxygen desaturation which may be difficult to appreciate clinically. At times, normal findings in the newborn may not exclude CHD. Confounding factors include duct-dependent pulmonary and systemic circulations. In addition, the initially high pulmonary vascular resistance restricts left to right shunts even when large communications between both circulations are present. Clinical findings may evolve over the first few days of the newborn's life as the duct closes, or over the next few weeks as the pulmonary vascular resistance drops thereby allowing for an increasing left to right shunt. This paper aims to alert the point of care clinician about the possibility of a cardiac abnormality and to suggest which may require expert assessment.

Keywords: *Neonatal; Congenital heart disease; Fetal echocardiography; Newborn pulse oximetry*

Introduction

Congenital heart disease (CHD) is the most common birth defect with a prevalence of approximately 1% [1-3]. Most abnormalities are mild, such as small ventricular septal defects (VSDs), and of little consequence. Some are more severe, such as transposition of the great arteries (TGA), which require immediate diagnosis and intervention to prevent short and long-term sequelae. A few abnormalities, such as an interrupted aortic arch, may lead to death once the ductus closes. Serious CHD requires specialist cardiology input and accounts for between 2.5-3/1000 live births [3]. For parents, the question as to whether there is something wrong with their baby's heart is especially concerning. They are aware that a cardiac abnormality is so small an infant may result in their baby dying. If serious, a timely diagnosis and appropriate management are required. What then are the clinical clues that suggest CHD?

Fetal Ultrasound Screening

Fetal ultrasound screening has significantly improved the prenatal diagnosis of CHD [4]. Widely practiced in Australia (over 96% of Victorian women [5]) and much of the developed world, it helps guide the clinician as to where to have the baby [6]. Specifically, a fetus diagnosed with duct-dependent circulation should be delivered at a tertiary center as he or she may require the infusion of prostaglandin E1 (PGE1) to maintain duct patency before surgical or catheter intervention. In utero transfer is much safer than post-natal newborn transfer [7]. In some centers, intrauterine cardiac interventions may be possible to hopefully optimize the baby's subsequent treatment [8]. For example, valvuloplasty for a narrowed aortic valve may improve the growth of the left ventricle [9]. One pooled study placed the sensitivity and specificity of the early fetal ultrasound for detecting severe CHD at 78.6% and 98.9% respectively [10]. When used as a screening tool in the second trimester, the overall detection rate of CHD by fetal

ultrasound has been quoted as up to 45.1% [11].

Fetal arrhythmias may generally be controlled by treating the mother which allows for the placental transfer of the medication to the fetus. For example, digoxin is prescribed to the mother to revert supraventricular tachycardia in the fetus [12]. Fetal echocardiography can monitor the left ventricular size and function if the tachycardia is recurrent or persistent, thereby guiding the time of the delivery.

Despite significant improvements in the diagnostic yield over the last decade, fetal echocardiography still has limitations. Firstly, the procedure is largely operator-dependent. Initial scans are generally performed by technicians, the detection rates ultimately being dictated by the skill and experience of the operator [13]. Secondly, other factors may interfere with the clarity of the images obtained, namely the position of the baby, habitus of the mother, and any scarring from previous abdominal surgery [14]. Thirdly, specific cardiac abnormalities may be difficult to detect. Not uncommonly they include some VSDs, coarctation of the aorta, TGA, and total anomalous pulmonary venous drainage (TAPVD) [10]. In utero, the ductus is large and shunts right to left, and may mask a localized coarctation. Other findings such as a dilated right ventricle or hypoplastic aortic arch may suggest a possible coarctation [15]. TGA with an intact ventricular septum may be missed because of difficulty in obtaining the "three-vessel view" needed to determine the great vessel connections, the intracardiac anatomy being normal [11,16]. Some abnormalities may evolve as the pregnancy advances, e.g., hypoplasia of the left ventricle from a stenotic aortic or mitral valve, which may be missed unless the scan is repeated in the third trimester [17].

Clinical Features

A full examination of the newborn may allow one to consider the possibility of a cardiac lesion if there are one or more of the following signs.

1. Murmurs

A murmur usually points to a cardiac abnormality. However, some murmurs may be related to flow across the cardiac structures. Such murmurs are generally soft with the rest of the cardiac findings being normal [11]. Occasionally a soft murmur may arise from a trivial CHD such as turbulence around a small VSD which often closes spontaneously, or from transient tricuspid regurgitation in babies with respiratory distress [11]. A moderately loud or loud murmur with a thrill suggests CHD which may be significant. For example, that arising from severe pulmonary stenosis in a baby with Fallot's tetralogy. Murmurs arising from communications between the systemic and pulmonary circulations (e.g., VSD) typically take days to weeks to develop as they require a fall in the pulmonary vascular resistance (PVR) to allow for an increased left to right shunt (see below).

It is especially helpful to recognize signs suggestive of pulmonary hypertension in the newborn particularly if there is no associated murmur. As the heart rate is rapid there is usually the summation of the second heart sounds (S2). Pulmonary hypertension should be suspected then if the summated S2 in the pulmonary area is louder than the S2 in the aortic area [11]. Conditions causing pulmonary hypertension include an interrupted arch or a tight coarctation [18].

2. Cyanosis

Cyanosis in the newborn requires an urgent diagnosis. In a duct-dependent pulmonary circulation, duct closure can result in increasing cyanosis which may lead to cerebral hypoxia. Such neonates require a definitive diagnosis which usually necessitates transfer to a tertiary center. PGE1 infusion is required to maintain duct patency for safe transfer. Cyanotic heart disease normally presents early, often within the first 24 hours of life. Central cyanosis is best assessed by viewing the mucous membranes. However, not all blue babies have cyanotic heart disease. Most relate to respiratory rather than cardiac causes [19]. Non-cardiac causes include upper airway obstruction (e.g., bilateral choanal atresia) or bruising of the face from a precipitate delivery [19]. Polycythaemia gives the baby a "ruddy blue" appearance. A high haematocrit and normal oxygen saturation help confirm the diagnosis. Murmurs may be associated with cyanosis (e.g., pulmonary stenosis in TOF) but maybe absent (e.g., TGA with intact ventricular septum).

Where the cause of cyanosis is unclear, Shinebourne's hyperoxaemic test can help to determine if it is cardiac or respiratory in origin. The arterial blood gas (pO2) is measured in room air and then on 100% O220, with a substantial rise if the cause is respiratory and a limited response if cardiac in origin (see table 1).

Condition	Suggested by
Primary lung disorder	A rise above 150mmHg on 100% O_2
Separate circulations Transposition of the great arteries	pO_2 20-35mmHg and a slight rise on 100% O_2
Right to left shunts Right ventricular outflow obstruction Hypoplastic left heart syndrome	pO_2 35-45mmHg with a rise of 10-20mmHg
Obligatory mixing Unobstructed total anomalous pulmonary venous drainage Truncus arteriosus	pO_2 60-70mmHg rising to 90s or even low 100s - the oxygen saturation will be 100%

Table 1: Shinebourne's hyperoxaemic test, results and interpretation [20].

3. Tachypnoea

Early tachypnoea (day 1-2 after birth) is generally non-cardiac and is most likely to be respiratory in origin. Rarely, it can be due to a metabolic cause [20]. Occasionally, it may be associated with cardiac failure, e.g., if there is a large systemic arteriovenous (AV) fistula where the left to right shunting is unaffected by the high PVR (e.g., an AV fistula of the great vein of Galen). Left ventricular "pump failure", such as arising from a cardiomyopathy or myocarditis may also lead to early tachypnoea. Tachypnoea from large left to right shunts as seen in large VSDs take a week or more to develop, depending on how quickly the PVR falls. Tachypnoea may occur early if there is a coexisting left-sided obstructive lesion e.g., hypoplastic left heart syndrome or congenital mitral stenosis.

4. Pulses

It is essential to feel the femoral pulses in all newborn babies. Occasionally, they are difficult to feel in the first 24-48h even if the baby's lower limbs are relaxed. If one is considering the possibility of a left-sided obstructive lesion, it is important to feel not only the brachial pulse (the radial pulse is often difficult to feel in a newborn) and the femoral pulse but also the carotids (see below). A weak pulse may imply a low cardiac output related to pump failure for example from a tight coarctation of the aorta, or critical aortic stenosis with a closing duct, or from a cardiomyopathy, myocarditis, or as often is the case, sepsis [21]. Bounding pulses may suggest aortic runoff from a large duct that fails to close.

5. Heart rate

The normal heart rate of a newborn is about 100-160bpm. Bradycardia of 60 or less should prompt an ECG to exclude complete heart block. Bradycardia of 70-90bpm may occur if the neonate is cold, be it due to sinus bradycardia or occasionally from non-conducted atrial ectopics. Rarely, it can be an indicator of long QT syndrome [11]. Tachycardias greater than 200bpm may suggest a supraventricular tachycardia, best picked up by a formal ECG. Irregular heart rates, not uncommon in the newborn, can arise from atrial ectopy which is usually transient [11].

6. Additional clinical signs

Hepatomegaly and peripheral oedema may be present if right heart failure develops, for example from severe pulmonary stenosis or secondary to a left-sided obstructive lesion or a large left to right shunt. Liver enlargement is best determined by percussion to assess the liver span. The hepatomegaly from cardiac failure tends to have a rounded edge, rather than a sharp edge when the lever is pushed down from a hyperexpanded lung [11], for example as seen in bronchiolitis. Peripheral oedema is uncommon and in infants requires firm pressure for thirty to sixty seconds to get a slight depression, akin to what is required when assessing lymphoedema.

7. Pulse oximetry

Pulse oximetry further aids the assessment of the newborn. It is an extremely useful screening tool, compulsory in some centers, with a sensitivity of approximately 76%. It may detect many neonates with serious CHD [20]. Pre- and post-ductal saturations are measured in the right hand and then a foot. A lower post-ductal reading may suggest the diagnosis of duct-dependent systemic circulation (see later) [22]. A normal (negative) result occurs if the PaO2 is >95% in both right upper and lower limbs or there is a <3% absolute difference in PaO220. Nevertheless, pulse oximetry may still miss significant duct-dependent lesions such as coarctation of the aorta or an interrupted aortic arch. Mild cyanosis, in the high 80s or low 90s, often difficult to pick up clinically, may point to cyanotic CHD such as an unobstructed TAPVD. It may also indicate mild desaturation from an ongoing respiratory problem [20].

Confounding Factors in the Newborn

1. Duct-dependent circulation

The presence of a duct-dependent pulmonary or systemic circulation may mask a serious congenital heart abnormality.

Duct-dependent pulmonary circulations

A large duct, for example, may improve the oxygenation in a baby with significant cyanotic heart disease making the desaturation is difficult to detect clinically. As noted above, saturations in the high 80s and low 90s may be difficult to pick clinically. Pulse oximetry resulting in a low reading may draw attention to the possibility of a cyanotic CHD e.g. an unobstructed TAPVD or even TGA when there is a large ductus and a reasonable sized atrial defect [20]. Cyanosis however will become more marked as the duct closes. When cyanosis is diagnosed, it may be due to causes other than cyanotic heart disease. Of importance is cyanosis that may occur from persistent foetal circulation (PFC) sometimes called persistent pulmonary hypertension, which may result from various causes such as sepsis, dehydration, meconium aspiration, and/or a hypoplastic lung from a diaphragmatic hernia [23]. All may cause the PVR to remain high or increase following an initial drop. As a result, there may be bidirectional shunting at the atrial level and right to left shunting at duct level. Cyanosis may ensue with a further reduction in the post-ductal saturations. An echocardiogram will be helpful as it will show a pure right to left shunt at atrial level with TAPVD compared to bidirectional shunting in PFC [24,25], in addition to determining the anatomy of the pulmonary venous return.

Duct-dependent systemic circulation

The right ventricle may help support the systemic circulation by shunting right to left through the ductus in such conditions as critical aortic stenosis or an interrupted aortic arch. It is therefore

important to feel all the pulses of the newborn if a significant left-sided congenital abnormality is suspected – the brachialis, femorals, and carotids [20]. At times however the femoral pulse volume will be better than the brachialis if the duct is wide open. Differential pre- and post-ductal saturations will be helpful in such cases. Gradual duct closure generally leads to the baby becoming somewhat unwell, with poor feeding, "blotchiness" and increasing tachypnoea. A differential pulse volume may then become apparent. Rapid duct closure may result in the baby presenting in a collapsed state which is often misinterpreted as due to an overwhelming infection [20]. As most babies are discharged on day two or three, such an occurrence may happen at home.

2. Changes in pulmonary vascular resistance (PVR)

The initial high neonatal PVR restricts left to right shunts even when there are large communications between both circulations. The reduction in PVR after birth is multifactorial. A rapid decrease is observed initially as inspiration expands the alveoli and capillary beds. A subsequent slower fall occurs as fluid is cleared from the lungs in the first hours after birth. New lung tissue is laid down in the first few weeks after birth, with an additional drop in the PVR aided by the pulmonary arteries losing their heavy musculature following a reduction in pulmonary artery pressures.

The changes in the PVR may delay the diagnosis of communications between the systemic and pulmonary circulations, for example, from a large VSD. As the PVR falls the shunting increases resulting in an audible murmur. This may take a few weeks and may only be apparent after the baby is discharged from the hospital. An ASD may take even longer to manifest as most atrial shunting occurs in diastole and depends not only on a drop-in PVR but also on increased compliance of the right ventricle as it atrophies.

It is therefore not possible to clinically exclude all CHD when the baby is discharged home with the mother. Review at about one month of age with the absence of a murmur at that time would tend to exclude most CHD remembering that an ASD or partial anomalous pulmonary drainage may take even longer to manifest clinically.

In Summary

A prenatal diagnosis of a potentially duct-dependent abnormality warrants delivery at a tertiary center with a newborn intensive care unit as the baby is likely to require a PGE1 infusion to maintain the circulation until surgical and/or other interventions are undertaken. A newborn baby that is clinically well with a soft murmur, without hemodynamic changes, who is acyanotic and with normal pulses can be further assessed at a convenient time. If cyanosed, an urgent diagnosis is required as the neonate may have a duct-dependent cardiac abnormality. If the duct closes, there will be increasing hypoxia and cyanosis with both short- and long-term sequelae. Early referral to a tertiary center for a definitive diagnosis is recommended, best done on a PGE1 infusion. Consider a duct-dependent systemic circulation in any baby with differential pulses, differential cyanosis measured on oximetry, or if the baby goes into a state of shock, which may not be due to sepsis. Moderate to loud murmurs suggest CHD while tachypnoea and/or differential or poor pulses may point towards a more serious cardiac abnormality. A review of the baby at one month of age may allow for the diagnosis of left to right shunts arising from communications between the pulmonary and systemic circulations. Any neonate requiring urgent surgical intervention may need an echocardiogram to exclude significant CHD which may not be clinically obvious at the time.

Conclusion

CHD is common and although most abnormalities are mild, serious lesions can be life-threatening requiring immediate diagnosis and treatment. Many complex congenital heart abnormalities are diagnosed prenatally. However, others are picked up in the newborn period by the presence of a significant murmur, tachypnoea, cyanosis, and/or differential pulses. Any suggestion of a duct-dependent circulation requires transfer to a tertiary center on PGE1 for definitive diagnosis and timely management. Pulse oximetry may be helpful in the assessment of the newborn before discharge. Finally, although the infant may appear well at birth, CHD may present later, suggesting the need for review following discharge from the hospital.

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