Evaluation of suspected congenital heart disease

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Abstract

Congenital heart disease (CHD) is the most common congenital anomaly in the United Kingdom (UK). Despite major advances in diagnosis and management over the last decade, CHD remains a leading cause of infant morbidity and mortality. Current existing screening tools fail to identify up to 50% of children with CHD before discharge from hospital. Pulse oximetry screening has been well studied and seems a promising screening tool. Combined use of pulse oximetry with fetal anomaly screening and routine newborn examination could detect the majority of critical CHDs in asymptomatic infants. Notably pulse oximetry screening has yet to be incorporated as part of universal screening programme in the United Kingdom. Echocardiography remains the gold standard investigation but is not always available. Chest X-rays and four limb blood pressures are of limited diagnostic value while electrocardiogram helps in only few diagnoses during infancy. A detailed history and thorough clinical examination are of paramount significance in suspected CHD in infants. As the clinical presentation can mimic or overlap with other common conditions in infancy (sepsis, respiratory or metabolic condition), a high index of suspicion with a systematic approach is vital for the timely diagnosis and management.

Keywords congenital heart disease; heart murmur; infant; neonate

Introduction

Congenital heart disease (CHD) is defined as defects in the heart and major blood vessels, including structural, chromosomal, genetic, biochemical defects and malformations. CHD is the commonest of all congenital anomalies seen in live births. The incidence of CHD in general population is about 1%, more precisely 6–8 per 1000 live births and the incidence of serious CHD (needing intervention or operation within 1 month after birth) is 1.6–2 per 1000. CHD could also be responsible for a number of spontaneous abortions, still births and medical termination of pregnancy, and exact incidence of which remains unknown.

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Major developments in diagnosis and management over the last decade have led to dramatic improvements in survival with more than 85% of children diagnosed with CHD now surviving into adulthood. Despite these advances, CHD remains a leading cause of infant mortality accounting for up to 40% of all deaths from congenital defects. 3–7.5% of the infant deaths in the developed world are reported to be due to CHD.

Current screening tests consisting of fetal anomaly screening (FAS) at 18–20 weeks of gestation and routine examination of newborn fail to detect a large proportion of CHDs. It is estimated that up to 50% infants with critical CHDs are discharged home undiagnosed. These conditions carry a significant risk of sudden collapse or even death before surgical intervention. Early diagnosis and timely appropriate management are therefore essential to improve outcome.

Aetiology

The aetiology of CHD is known to be multi-factorial. These include genetic predisposition and environmental stimuli. A family history in close relatives increases the chances of CHD in subsequent pregnancy. A mother with CHD has 6% risk of having an affected offspring while an affected father has a 2% risk. Having a child with CHD confers a risk of 2–3% in subsequent pregnancy but having a child with hypoplastic left heart syndrome (HLHS) or having two or more children with CHD could increase the risk of CHD by 10% in a subsequent child, a roughly ten-fold increase in risk.

There has been significant progress in understanding the genetic pathogenesis of CHD in children. A recent study reported that *de novo* point mutations in several hundred genes collectively contribute to approximately 10% of severe CHDs. Environmental factors during critical developmental window might result in similar effects. The proportion of CHD associated with chromosomal anomalies varies between 4 and 12% but can be as high as 22% if antenatal deaths in fetuses are included. Certain CHDs have strong association with specific chromosomal defects such as aortic arch anomalies and 22q11 deletion.

Although hereditary and genetic factors play a major role in cardiac malformations, some causative teratogens are identified (see Table 1).

There are some common associations between CHD and other conditions in the child. There is a significant increase in risk of CHD in children with arrhythmias and prolonged QT syndrome. CHD can also be a part of a syndrome such as Cornelia de Lange syndrome (VSD), Holt–Oram syndrome (ASD, VSD) and CHARGE association (Fallot's tetralogy, truncus arteriosus and aortic arch anomalies). CHD is frequently seen in connective tissue disorders such as Marfan syndrome (aortic dilatation, aortic aneurysm formation, mitral valve prolapse and regurgitation, aortic regurgitation) and Ehlers–Danlos syndrome (aortic dilatation and aneurysmal formation, ASD).

Antenatal screening and fetal echocardiography

Most babies with cardiac defects are born to mothers with low or no identifiable risk factors for CHD, emphasizing the importance of basic fetal echocardiography as part of routine anomaly screening. In the UK, FAS is universally offered at 18–20 weeks of gestation or earlier in high-risk cases. Any suspicion of CHD

Aetiology and associations in congenital cardiac defects

Association	Causation	Cardiac defects
Genetic		
Chromosomal anomalies	Trisomy 21	AVSD, VSD, ASD, TOF
	22q11 deletion	Aortic arch defects, TOF
	Trisomy 18	VSD, ASD, PDA
	Trisomy 13	VSD
	Alagille syndrome (microdeletion on chromosome 12, autosomal dominant inheritance)	Peripheral pulmonary artery stenosis
Single gene defects	Noonan's syndrome	PS, HOCM, ASD
	Williams syndrome	Supra valvular AS, peripheral PS
	Turner syndrome	CoA, BAV, AS
Maternal condition		
	Maternal diabetes	Cardiomyopathy, VSD, PDA, TGA
	SLE, connective tissue disorder	Congenital heart blocks
Environmental		
Maternal alcohol abuse	Fetal alcohol syndrome	VSD, ASD, PDA
Maternal rubella	Congenital rubella syndrome	PS, PDA
Maternal lithium ingestion	Ebstein's anomaly	Hypoplastic right ventricle
Maternal phenytoin ingestion	Multiple defects	PS, AS, COA, PDA
Maternal amphentamines ingestion	Multiple defects	ASD, VSD, TGA, PDA
Maternal valproic acid ingestion	Multiple defects	ASD, VSD, AS, PA, COA
Maternal retinoic acid ingestion	Great vessels defect	Conotruncal anomalies

Table 1

on screening or high risk factors in women warrants specialist fetal echocardiography within 1–2 weeks.

Evidence suggests that delivery of antenatally diagnosed HLHS or transposition of great arteries (TGA) in or close to a specialist paediatric cardiology surgical centre improves their survival and neurodevelopmental outcome. Whilst there is no such evidence for other types of CHDs early antenatal diagnosis helps in planning perinatal management, counselling parents and allowing anticipation of problems. The detection of other associated congenital deformities will allow parents to prepare mentally and emotionally for their child's likely course of events during pregnancy or after birth. The indications for fetal echocardiography are summarised in Table 2.

The overall antenatal detection rate of CHD remains low averaging 30–50% in the UK. Although some tertiary specialist screening centres report higher rates of diagnostic accuracy, the reported detection rate varies significantly (3–70%) depending on the expertise of ultrasonographers and the type of CHD. Currently, various international bodies are working on formalising and auditing the standards of FAS teaching and training in order to improve antenatal diagnosis rate.

Pulse oximetry screening (POS)

Pulse oximetry screening for CHD has been the subject of several well-designed large randomised controlled trials. In conjunction with pre-existing screening tools, POS can detect up to 78–92% of critical CHDs.

The authors from a recent systematic review and meta-analysis (2012) involving over 230 000 children concluded that POS has high specificity (more than 99–99.6%) and moderate sensitivity (75–80%) in detecting critical CHD in asymptomatic neonates. POS has also been reported to be feasible and cost-effective based on current clinical settings. POS was shown to be well tolerated, simple, and acceptable to parents and clinical staff. These findings are substantial enough to meet the criteria for universal screening.

In 2011, the US Secretary's Advisory Committee in Heritable Disorders in Newborns and Children convened a workgroup which recommended a standard protocol for routine screening. The subsequent statement by this group was endorsed by a number of professional bodies, including the American Academy of Paediatrics, American Heart Association and the US Health and Human Services Secretary. States across the USA are currently considering implementation of this recommendation. To date, four are screening all neonates, with the majority of others making progress towards this goal. In Switzerland and Sweden, most neonates are screened as part of their routine postnatal care.

In the UK, the National Screening Committee has undertaken a consultation on the value of POS in diagnosing CHD. At the time of writing the conclusions of this review are awaited. A recent published study (2012) showed that only 36 of 204 units (18%) in the UK have been performing POS routinely, but each varied significantly in practice including the timing of screening, acceptable saturation threshold, and whether both pre- and postductal saturations were measured.

Clinical presentation

The signs and symptoms of CHD may be non-specific in infants. Box 1 summarises the signs and symptoms that suggest CHD.

Summary of indications for fetal echocardiography

Fetal factors:

- Suspected cardiac anomaly on FAS ultrasound
- Increased nuchal translucency thickness
- Abnormal fetal karyotype e.g., trisomies (21, 18, 13), Turner syndrome, 22q11 deletion
- Other congenital anomalies like exomphalos, diaphragmatic hernia etc
- Fetal hydrops
- Fetal arrhythmia
- Twin to twin transfusion syndrome

Maternal factors:

- Increased maternal risk for Down's syndrome and other congenital defects (advanced maternal age or increased risk of Down's syndrome on serum screening)
- Maternal alcohol abuse
- Maternal ingestion of teratogenic drugs (anticonvulsants, lithium, retinoic acid, amphetamines)
- Maternal diseases (insulin dependent diabetes mellitus, phenylketonuria, exposure to Rubella, Coxsakie or Parvo virus infection, maternal Ro/La antibodies)

Familial factors:

- Family history of CHD in first degree relative
- Genetic syndromes

Table 2

The clinical presentation of CHD conditions can be broadly divided into three categories:

- Cyanotic CHD presenting as blue baby
- CHD presenting with neonatal collapse or even sudden death in infancy
- CHD presenting as heart murmur or heart failure

Congenital heart defects presenting as the blue baby

Cyanotic congenital heart defects constitute about 20% of CHD. Most of the cyanotic CHDs in infancy start with 'T' (Box 2).

Pointers to CHD in infants

- Unwell infant
- Feeble or absent pulses (brachial or femoral)
- Persistent cyanosis in absence of respiratory distress or cyanotic episodes
- Low oxygen saturation (<95% in air) or difference of >2–3% between pre- and post-ductal saturations
- Heart murmur
- Presence of dysmorphic features or other congenital anomalies
- Collapse/sudden death
- Presence of arrhythmias or heart failure
- Suspicion of CHD on FAS
- Positive family history of CHD

Box 1

Cyanotic congenital heart diseases – five 'Ts' or rarely 'PAE'

- **T**ransposition of great arteries (TGA)
- **T**etralogy of Fallot's (TOF) and double outlet right ventricle (DORV)
- **T**runcus arteriosus
- **T**otal anomalous pulmonary venous connection (TAPVC)
- **T**ricuspid atresia
- **P**ulmonary atresia with no VSD and severe pulmonary stenosis (considered severe spectrum of tetralogy of Fallot's)
- **E**bstein anomaly (rare with variable presentation)

Box 2

The timing of presentation varies from soon after birth to late in infancy or childhood depending upon ductal physiology, pulmonary circulation dependency upon duct, degree of right ventricular outflow obstruction, and presence of other associated cardiac defects. TOF remains the most common cyanotic CHD while TGA is the most common cyanotic CHD presenting within the first week after birth. TOF presents with variable cyanosis depending on the severity of outflow tract obstruction and shunt across VSD.

Most babies with cyanotic CHD are *not* breathless except in obstructed TAPVC or late presenters with heart failure or circulatory compromise. Clinical detection of cyanosis is unreliable; which explains the vital role of POS. Oxygen saturations of less than 95% in air or a difference over 2–3% between pre- and post-ductal saturations, is considered significant. Obstructed TAPVC should always be considered in any infant with cyanosis and respiratory distress, which can be clinically difficult to differentiate from persistent pulmonary hypertension of the newborn (PPHN).

The chest X-ray is helpful in diagnosing respiratory pathology and differentiating types of CHD. Pulmonary oligaemia is seen in TOF, Ebstein anomaly, critical pulmonary stenosis and pulmonary atresia due to decreased pulmonary flow. Pulmonary plethora is seen in TGA with intact interventricular septum, truncus arteriosus, tricuspid atresia, TAPVC, DORV and single ventricle. The classical 'cardiac silhouette' on X-ray, although commonly thought to be diagnostic, can often be an unreliable finding. Massive cardiomegaly frequently indicates Ebstein anomaly. Where it is available paediatric echocardiogram is the investigation of choice.

Initial cardiorespiratory stabilisation, prostaglandin infusion and correction of any metabolic acidosis form the mainstay of management of cyanotic CHDs. TGA is generally repaired within 1–2 weeks after birth but some infants may need urgent atrial septostomy to improve central mixing. Primary surgical repair at 4–6 months is preferred for TOF. Some cyanotic heart conditions may need palliative procedure (like modified BT shunt) to improve pulmonary blood flow while waiting for corrective surgery.

Hypoxic spell

A hypoxic spell, also known as 'tet spell', 'cyanotic spell' or 'hypercyanotic spell', is not uncommon in children with TOF

awaiting surgery. The hypoxic spell is characterised by hyperpnoea, irritability and prolonged crying, increasing cyanosis, and the disappearance of any heart murmur. A severe episode may lead to collapse, convulsion, stroke or even death.

The mechanism of these hypoxic spells is related to the relative resistance in the pulmonary outflow tract and systemic vascular resistance (SVR). Due to the presence of a large VSD in TOF, pressures in both ventricles equalises and may be viewed as a single chamber that pumps blood to both systemic and pulmonary circulations. Hence either an increase in pulmonary vascular (PVR) resistance or a decrease in SVR will increase the degree of right-to-left shunt, causing arterial desaturations. Understanding this mechanism is the key in managing 'hypoxic spell' (Table 3).

Congenital heart defects presenting as neonatal collapse/ sudden death in infancy

Infants with duct-dependant CHDs such as coarctation of aorta (CoA), HLHS, interrupted aortic arch and critical aortic valve stenosis, may present as neonatal collapse or even sudden death if diagnosis or management is delayed.

The systemic circulation in these CHDs relies upon ductal patency. The ductal closure can lead to impaired lower or even whole body tissue perfusion leading to impalpable pulses and progressive acidosis, which explains the usual timing of presentation within first 2–3 weeks after birth. Many of these conditions, like CoA, can be difficult to diagnose antenatally. They typically remain asymptomatic and are notoriously difficult to detect during routine examination of newborn whilst the ductus arteriosus is widely open. Moreover, in these circumstances they may not be identified even on POS. Thus timely diagnosis can be challenging. When the ductus arteriosus begins to close they present with sudden collapse or shock.

Others follow a more insidious course of poor feeding, lethargy, breathlessness, hypothermia or cyanotic episodes mimicking other common neonatal conditions such as sepsis, respiratory and metabolic disorder. Presentation of poor feeding at this age, due to its non-specificity, is a frequently neglected symptom. These infants often present with heart failure or circulatory compromise. The clue to a cardiac cause would be weak or absent peripheral pulses and pre- and post-ductal saturation difference, although this may not be present in all cases (Table 4).

Whilst echocardiography is diagnostic it depends on the availability of paediatric cardiologist/paediatrician with expertise in cardiology. ECG and chest X-ray are of limited diagnostic value.

Management includes initial stabilisation, correction of metabolic acidosis (if any) and commencement of alprostadil/ dinoprostone while awaiting definitive surgical repair. There should not be a delay in starting alprostadil/dinoprostone while waiting for echocardiogram or cardiology opinion.

CHD presenting as heart murmur or heart failure

Almost all cardiac conditions can manifest with a heart murmur. In fact, heart murmur detected during routine examination is the commonest presentation in CHD. However, some serious CHD like TGA or TAPVC may not have a murmur. A murmur may often not be heard in babies who are examined soon after birth due as pulmonary vascular resistance is still high and there is therefore minimal shunting across VSDs or decreased pulmonary blood flow.

Heart murmur and the risk of having a CHD

Nearly 80% of children have a heart murmur at some stage during childhood. In majority especially between ages 1–6 years, the murmurs are usually non-pathological. Heart murmur can also be a common finding during transition circulation after birth.

However, murmur heard during early infancy carries a high risk of CHD association. Wren et al. reported around 25% of infants with murmurs heard at the 6-week check had structural heart defects while another study showed that over half of murmurs heard in neonates were pathological. Moreover, the life-threatening duct-dependent cardiac malformations often

Step-wise management of hypoxic spell

Initial steps:

- The infant should be held in a knee–chest position (increases SVR and decreases right-to-left shunt)
- Oxygen is usually administered but has little effect on oxygen saturation
- Morphine sulphate subcutaneously or intramuscularly (suppresses the respiratory centre and abolishes hyperpnoea, hence breaks the vicious circle)
- Acidosis should be treated with sodium bicarbonate intravenously (reduces the respiratory centre stimulant effect of acidosis)

If child remains un-responsive to above measure following drugs can be helpful:

- Beta-blockers (Propranolol or esmolol) administered by slow intravenous push (reduces the heart rate and may reverse the spell)
- Vasoconstrictors such as phenylephrine intravenously may be effective (by raising SVR)
- Ketamine can be effective (by increasing SVR resistance and abolishing respiratory drive by sedation effect)

Table 3

Characteristics of pulses in obstructive left heart lesions

Congenital heart defect	Nature of pulse
Hypoplastic left heart syndrome	Weak brachials and femorals
Critical aortic stenosis	Absent brachials and reasonable femorals
Interrupted aortic arch	Good right brachial pulse, absent left brachial pulse and weak femorals
Coarctation of aorta	Weak femorals, sometimes weak brachials

Table 4

manifest during this period of life. For reasons mentioned, murmur in this age group need to be assessed methodologically including a relevant history, thorough cardiovascular examination, appropriate investigations and early referral to paediatric specialists.

A simplified approach to evaluation and initial management of infants with suspected congenital heart disease

The clinical presentation of CHD can be non-specific mimicking other common conditions in infancy (sepsis, respiratory or metabolic condition). Therefore, having a high index of suspicion and systematic approach is vital for the timely diagnosis and management.

The initial evaluation of an infant with suspected CHD should include a detailed history including obstetric history and family history, meticulous physical examination (including palpation of peripheral pulses and measuring **pre- and post-ducal saturations**) and hyperoxia test. Careful examination for dysmorphic features and other congenital anomalies may reveal a clue to diagnosis.

A right-to-left shunt via PDA (from pulmonary artery to aorta) results in higher pre-ductal saturation than post-ductal saturation (differential cyanosis) which can be seen in PPHN and severe left heart obstruction (CoA, critical aortic stenosis, hypoplastic or interrupted aortic arch). Persistently reversed differential cyanosis (higher post than preductal saturations) is rare. It is sometimes seen in TGA with PPHN or coarctation. It can be noted transiently in PPHN or in well infants soon after birth.

The **Hyperoxia test** is particularly useful in differentiating the cardiac causes from respiratory causes in cyanotic newborns. It works on the assumption that when there exists a right-to-left shunt or central mixing in cyanotic CHDs, regardless of the level of alveolar oxygenation, the desaturating effect of the shunt will not alter. On confirmation of central cyanosis by measuring the arterial partial pressure of oxygen (PaO_2), response of PaO_2 to 100% oxygen inhalation is tested (hyperoxia test). Oxygen should be administered through a plastic hood for at least 10 minutes in order to fill the alveolar spaces completely with oxygen. In CHD, the rise in PaO_2 is usually no more than 10–30 mmHg and hardly ever exceeds 100 mmHg. With pulmonary diseases, PaO_2 often rises to over 100 mmHg.

However, infants with massive intra-pulmonary shunt from a respiratory disease may not show a rise in PaO_2 to 100 mmHg. Conversely, some infants with cyanotic defects with a large pulmonary blood flow, such as TAPVC, may demonstrate a rise in PaO_2 of 100 mmHg or higher. Therefore, hyperoxia test should be interpreted in the light of clinical picture and the degree of pulmonary pathology seen on X-ray.

Four limb blood pressures are of limited value in neonates but can be helpful in older children. If the systolic pressure in right arm exceeds 10 mmHg difference as measured in the leg, an aortic arch anomaly is likely. PDA may not allow this gradient to manifest. Therefore the absence of a systolic pressure gradient alone does not rule out arch anomaly.

ECG is of diagnostic value showing superior axis deviation in infants with tricuspid atresia, atrioventricular septal defect (AVSD) and congenitally corrected TGA (ccTGA).

Chest X-ray is important in excluding respiratory pathology. It allows examination of the lung fields for pulmonary vascularity which helps in differentiating type of CHD.

Echocardiogram, although not always readily available, remains the gold standard in diagnosing CHD. Initial management should not be delayed while waiting for an echocardiogram. Once the diagnosis of CHD is suspected, the priority should be to stabilise the infant before further arrangements for a definitive diagnosis.

Infants presenting with cardiorespiratory failure should first be stabilised as per Advanced Paediatric Life Support guidelines. Fluid resuscitation and inotropic support may be required. Antibiotic cover usually needed as sepsis is common and difficult to rule out in this age group. Infants with cardiorespiratory failure often will need intubation and ventilation, which should be done under adequate sedation, analgesia and paralysis to avoid serious arrhythmias, bradycardia or even asystole.

Maintaining ductal patency can be lifesaving in infants with suspected duct-dependent heart condition and there should not be a delay in starting alprostadil/dinoprostone while awaiting echocardiogram or specialist advice. A small dose (5–10 ng/kg/minutes) is normally required to maintain ductal patency in babies with open 'duct' (antenatal diagnosis or early presentation). Collapsed infants with weak or absent femoral pulses may need higher dose, up to 100 ng/kg/minutes. The desired response would be an improvement in oxygen saturation in cyanotic CHD, and improvement in acidosis, lactate or femoral pulses in collapsed acyanotic infant. The dose can be doubled if response is inadequate, and progress should be reassessed every 20–30 minutes. These infants should be discussed with paediatric cardiologists at the first opportunity.

Infants on low dose of alprostadil/dinoprostone (less than 15 ng/kg/minutes) are unlikely to have apnoea and do not routinely need mechanical ventilation. A 10-year retrospective population based audit conducted by the New South Wales transport service in Australia and a UK study reported safe transfer of infants with low dose alprostadil/dinoprostone without mechanical ventilation. As prostaglandin infusion can cause apnoea and hypotension, continuous cardiorespiratory monitoring is indicated.

Failure to respond prostaglandin could mean incorrect diagnosis, insufficient central mixing despite ductal patency (such as in TGA with restrictive atrial septum) or a lack of ductal response to prostaglandin. TGA with restrictive atrial septum is a time critical emergency. The infant is at great risk of sudden collapse or even death without atrial septostomy. Urgent discussion with a paediatric cardiologist is needed in these situations.

The future

The future is likely to see continual advances in antenatal diagnosis of CHD with a greater proportion being detected during pregnancy. In conjunction with current screening tools, implementation of POS should improve detection of critical CHD in asymptomatic infants. Increasing availability of neonatologists and Paediatricians with Expertise in Cardiology (PEC) should facilitate timely diagnosis and intervention. Further development of regional cardiology networks and shared care pathways should facilitate in delivering high-quality patient care near home. Increasing availability of telemedicine should also help with early diagnosis and prevent inappropriate transfers.

Conclusion

Despite major advancement in screening tools, CHD continues to be a diagnostic challenge in the neonatal period and infancy. Pulse oximetry screening, used in conjunction with fetal anomaly screening and routine examination of newborn, is helpful in detecting critical CHDs. A stepwise approach focussing on detailed history, meticulous physical examination and appropriate investigations should allow one to diagnose and manage suspected CHD appropriately even without immediate access to echocardiography facility. ◆

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Practice points

- Antenatal screening is a useful tool in detecting major CHD allowing better planned management.
- Life-threatening cardiac defects are often asymptomatic at birth and should be part of differential diagnosis in any unwell infant. High index of suspicion and systematic approach is vital for early diagnosis of CHD.
- Clinical presentation is often non-specific mimicking other common neonatal conditions like sepsis, respiratory pathology and metabolic disorders.
- Detailed history and meticulous physical examination, including palpation of brachial and femoral pulses, are essential. Looking for dysmorphic features and other anomalies can aid in diagnosis while obstetric history and family history of CHD can give a clue to underlying condition.
- Clinical detection of cyanosis is unreliable; hence pre- and post-ductal oxygen saturation should always be measured in all infants suspected to have CHD.
- Echocardiogram is the gold standard investigation but not mandatory in the initial evaluation (treatment should not be delayed while waiting for an echocardiogram).
- Commencement of Prostaglandin can be lifesaving intervention while waiting for a definitive diagnosis.
- Karyotype and/or specific genetic tests should be carefully considered and arranged.