Early Detection of Congenital Heart Disease

Introduction

Congenital heart disease (CHD) is one of the commonest human malformations, affecting 6 per 1000 livebirths. They account for 10% of infant deaths and about 50% of deaths from malformations. It is a clinical paradox that the most benign lesions such as small VSDs or mild pulmonary stenosis, are more likely to be detected on routine newborn examination. Of the major structural lesions, the cyanotic conditions will usually present with symptoms or signs and so also will be detected early. There remains however an important group with major structural lesions, particularly those with ductal dependent systemic circulations, who are well in the period shortly after birth and then collapse in a critical state once the ductus closes. In an era of early obstetric discharge, this collapse is increasingly likely to happen at home where successful resuscitation is more difficult. This clinical problem is one of the major unresolved challenges of newborn screening.

Epidemiology

The population based studies from the North of England provide the most rigorous insight into the limitations of routine newborn examination. They found that the newborn examination was normal in 55% of babies who were later diagnosed with CHD. In their population, only a third of babies with abnormal examinations were referred for early diagnosis. The rate of abnormal newborn examination varied from 0% with total anomalous pulmonary venous drainage to 75% of babies with pulmonary stenosis. Of the whole cohort with CHD, 54% were still undiagnosed at 6 weeks and 36% by 12 weeks.

Of most concern, 21 out of 1227 died before a diagnosis had been made. The diagnosis most likely to cause death before diagnosis were the left ventricular outflow tract obstructions, including hypoplastic left heart syndrome, critical coarctation, interrupted aortic arch and aortic stenosis. The same group studied a cohort of 120 babies with one of these 4 diagnoses. About a third of these babies had had an abnormal newborn examination but only half of these were referred for early diagnosis meaning 78% of these babies had left hospital before a diagnosis had been made. Twelve of these babies died within 24 hrs of birth but a further nine died between hospital discharge and diagnosis.

Risk Factors

These include:

- **Family History**: One previous sibling with CHD increases the recurrence risk to 2%, two previous siblings increases the risk to 10%.
- **Maternal Diabetes**: This is associated with an increased risk to 2-3%. Whether this is true in an era of better diabetes control is not known.
- **Other fetal abnormalities** on prenatal screening, including malformations of other systems, fetal arrhythmia's and non-immune hydrops.
- ** Syndromes and other structural malformations** diagnosed postnatally. Any baby with dysmorphic features or a structural malformation should be considered as at high risk for CHD. CHD is a common in most chromosomal abnormalities and in many other non chromosomal syndromes, including fetal alcohol syndrome and congenital infections such as rubella.
• **Down Syndrome** is the commonest trisomy and 35-45% will have CHD. Tubman et al\(^7\) found that only 56% of 27 Down's babies with CHD had an abnormal newborn physical examination. The same authors found chest xray and ECG could not be relied on to make the diagnosis accurately. Wren et al\(^2\) found that 25% of Downs babies with total atrioventricular canal defect had not had the diagnosis made by 12 weeks of age. Most would now recommend routine early echocardiography.

### Diagnosis.

#### Antenatal Diagnosis:

Most babies within the Australian health care system will have had an ultrasound screening for malformations. This examination usually includes a four chamber heart view. While there is evidence that with appropriate training, the four chamber view could detect 67% of all CHD, the reality in 1999 in the UK was that only 23% of major CHD was being detected before birth.\(^8\) The use of the four chamber view also influences the type of lesions detected antenatally. Conditions with a major impact on chamber size such as the hypoplastic left heart syndrome are more likely to be detected while conditions with little impact on chamber size such as transposition or coarctation are less likely to be detected. Detection rates in high risk pregnancies are much higher than for general screening and reflect the fact that referral is usually made to a centre with expertise in fetal echocardiography.

#### Postnatal Diagnosis:

Congenital heart disease presents in one of four ways in the newborn period.

- An abnormal examination in an asymptomatic baby, usually a murmur.
- Cyanosis.
- Heart failure / respiratory distress.
- Shock / cardiovascular collapse.

### Abnormal clinical examination in an asymptomatic baby.

Routine newborn examination of the cardiovascular system is not just auscultation and should include:

- Assessment of colour.
- Assessment of peripheral pulses including femorals.
- Assessment of the praecordial impulses.
- Auscultation for normal heart sounds and murmurs.

**Assessment of colour.** Many babies with cyanotic heart disease will be obviously blue from birth but some can be quite subtle, particularly with common mixing situations or ductal dependent pulmonary circulations before ductal constriction. This is further confused by babies often having blue lips and extremities in the first day or two after birth. Looking at the tongue can be helpful in babies with blue lips but, if in doubt, put an oxygen saturation monitor on the baby's foot.

**Assessment of peripheral pulses.** In post-ductal coarctation or preductal coarctation with a constricting duct, weak femoral pulses may be the only clinical pointer. Unfortunately in the latter condition, while the duct is still patent, the femorals may be easily palpable. Feeling the femorals can be difficult in a vigourous baby. One useful technique is to hold the leg in your hand and run your thumb up the medial border of the quadriceps muscle, your thumb will usually fall on the femoral pulse while your hand can limit the leg movement.

**Assessment of praecordial impulses.** While there is no rigorous study of the accuracy of this sign, many major CDH lesions will produce an increased right and/or left ventricular load well before a murmur appears. So an easily palpable or visible praecordial impulse in a quiet baby should always be taken seriously as a possible sign of CHD.
Murmurs. This is the commonest and the best studied of the physical signs. In two larger studies, murmurs were detected in 0.6% and 0.9% of routine newborn examinations. Benson et al\textsuperscript{9} reported 38% of these had a structural heart lesion while Ainsworth et al\textsuperscript{10} found CHD in 54%. In the latter study this represented a sensitivity of 44% and a positive predictive value of 54% for a murmur detecting CHD. On the other hand, the finding of a murmur increases the risk of CHD from 0.6% to about 40%. Of the innocent murmurs, most are due to patent ductus arteriosus or left pulmonary artery flow murmurs.\textsuperscript{11} The former usually close and the latter have usually resolved by 6 months. Of the babies with a murmur and CHD, most have a VSD and in our experience these are usually, but not always small muscular VSDs. But in Ainsworth series, 10 of 25 had major CHD including coarctation, aortic stenosis and Fallot's tetralogy.

![Image](image1.png)

**Figure 1: Shows the colour Doppler of a small perimembranous VSD.**

At a practical level, differentiating between an innocent and significant murmur can be difficult and the above data would suggest all newborn babies with murmurs should be investigated. However the following clinical pointers suggest that a murmur may be significant:

- **Other abnormal cardiac signs** as detailed above.
- **Loudness and radiation.** If a murmur is easy to hear and particularly if it radiates to other areas, then it will often be significant. If you're struggling to hear it in one area then it's more likely to be a transitional innocent murmur.
- **Persistence.** Transitional murmurs are just that and often go away, even within the space of a day. If a murmur persists, it should be investigated however soft it is.
- **Abnormal heart sounds.** Likewise if the heart sounds aren't normal, a murmur should always be investigated. The commonest abnormal heart sound in CHD is a loud S2.

**Cyanosis**

CHD is only one of several conditions that can present with cyanosis. The differential should include:

- Respiratory problems.
- Apnoea
- Seizures
- Methaemoglobinemia (very rare but always remembered)

While babies with cyanotic heart disease may present blue at birth, in many it is less obvious. The oxygen saturation has to be below 80% before cyanosis becomes clinically obvious. In some cyanotic conditions, the resting saturations will be in the 80's and so will not be obvious. Conditions that do this include anything where there is common mixing (transposition with VSD, unobstructed TAPVD, single ventricles) or ductal dependent conditions while the duct is patent (tricuspid or pulmonary atresia). Ductal dependent lesions eventually present with rapidly progressive cyanosis as the duct closes but the mixing situations may take some time to present. If there is any doubt about a baby, put a saturation monitor on the foot. If it is persistently less than 90%, further investigations should be intitiated.

The commonest condition presenting with neonatal cyanosis is transposition of the great vessels but any cyanotic CHD lesions can present in the newborn period.
Heart Failure / Respiratory distress

It is unusual for the common left to right shunts to present in the newborn period because pulmonary pressures take longer to fall in the presence of a large left to right shunts. Typically, large VSDs don't produce symptoms or signs in the neonatal period but present in failure at 2 to 4 weeks of age after the pulmonary pressures have fallen. Occasionally babies with left to right shunts will present with respiratory signs (usually tachypnoea), particularly if there is shunting at more than one site i.e. VSD and PDA or patent foramen ovale.

Major CHD can manifest as severe respiratory distress with high oxygen requirements and can closely mimic primary respiratory disease and or PPHN. Skinner reported a series of 34 such babies referred for assessment of PPHN of whom one had CHD.\(^{12}\)

Obstructed total anomalous pulmonary venous drainage is the classic condition that does this. In this condition, the pulmonary veins drain to systemic veins or the right atrium. If this drainage is obstructed, pulmonary venous congestion and oedema results in a respiratory clinical presentation. This condition is also one of the easiest to miss on echocardiography because the intracardiac anatomy is often normal. Transposition of the great arteries and coarctation of the aorta are the other conditions that can present this way.

The above highlights that any term baby with respiratory distress and persistent high oxygen requirements should have CHD excluded with an echocardiogram.

Shock /cardiovascular collapse.

This is the classic presentation of the ductal dependent obstructive left ventricle conditions (hypoplastic left heart, critical aortic stenosis and coarctation). In these conditions, the only way blood can reach the systemic circulation is via the ductus. They are often asymptomatic while the duct is patent and then collapse any time during the first week when it closes. They present pale and shocked with respiratory distress and weak pulses. The liver is invariably very large. They can be mistaken for a septic picture. Diagnosis is with echocardiography. Treatment depends on re-opening the duct with prostaglandin E2.

Investigation

Echocardiography.

This is the definitive test and in situations where there is early access to these skills, most of the other investigations become superfluous. Early echocardiography to exclude CHD should be arranged (see internal echocardiogram guideline) in the following situations.

- Any baby with signs suggestive of CHD (see above).
- Any baby with a persisting murmur on newborn examination.
- Any baby with severe hypoxic respiratory failure.
- Any baby with Down syndrome.
- Any baby with malformations that might be associated with CHD.

**Chest Xray and ECG.**

The traditional cardiac investigations of CXR and ECG are still useful in situations where there is limited access to echocardiography although both are of limited use in excluding heart disease. In older infants and children, Temmerman et al. found that a CXR helped in the diagnosis of heart disease in only 7% of cases. In babies with Down's syndrome, CXR had a sensitivity of 44% but specificity of 98% for CHD while ECG had a sensitivity of 41% and specificity of 100%. So if either of these tests are abnormal, they help, but if they're normal it doesn't exclude heart disease.

**Hyperoxia test (alias Nitrogen Washout).**

This is used for differentiating cardiac from non-cardiac causes of cyanosis. Most babies with cyanotic heart conditions will not increase pO2 significantly if placed in 100% oxygen. Babies are placed in 100% oxygen for 10 minutes, a preductal (either arm) arterial pO2 is measured. A pO2 over 150 mmHg makes cyanotic CHD unlikely. A pO2 under 150 mmHg is most likely to be CHD but severe respiratory problems and/or PPHN are not completely excluded.

**Upper and lower limb blood pressures.**

This may be useful in suspected coarctation although the accuracy of non-invasive blood pressure measures in babies is open to question. The difference between upper and lower limb blood pressures should be less than 15 mmHg.

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**Future Directions**

**Improving Antenatal Detection.**

There is a current focus on expanding the cardiac component of the 18 week anomaly scan to include a ventricular outflow view as well as a four chamber view. Even with this many cardiac lesions will continue to be missed.

**Oxygen Saturation Screening**

The major CHD lesions that are most likely to be missed in the newborn period will also have borderline low oxygen saturations in the lower limbs. In hypoplastic left heart or coarctation, the lower body blood supply is fed from the right ventricle via the ductus. In common mixing situations such as single ventricles or TGA with big VSD, venous and arterial blood mix before entering the systemic circulation. In TAPVD, the pulmonary and systemic venous return mix before passing right to left through the foramen ovale and duct. This fact has led some to explore the possibility of screening all newborn babies with a lower limb oxygen saturation in addition to the usual routine examination.

Several studies of this have now been completed and these studies have recently been the subject of two systematic reviews by Valmari and Thangaratinam et al. Thangaratinam's conclusion was that, although this is not a perfect screening test, if a baby does not have oxygen saturation screening, discharge of an apparently healthy baby with undiagnosed CHD is 5.5 times more likely for cyanotic CHD and 4.4 times more likely for all serious congenital heart disease. Thangaratinam et al concluded that summary estimate of accuracy were 63% sensitivity and 99.8% specificity and that this was a highly specific tool with a low false positive rate. One study assessed the added value of pulse oximetry to the clinical examination for detecting all CHD, with clinical examination detecting 46% and pulse oximetry detecting a further 31%, resulting in a combined sensitivity of 77% with 99.7% specificity. The addition of pulse
oximetry to newborn screening has the potential to diagnose CHD in an additional infant for approximately every 500 infants assessed for early screening (<24 hours) and every 2700 assessed for later screening, with a false positive rate of 0.2%.

The potential to prevent late presentations of critical CHD warrants the addition of oxygen saturation screening into the normal newborn examination of babies at RPA.

Residents: What to do?

- Examine all babies’ cardiovascular system thoroughly. Don't just focus on murmurs.
- If you find anything abnormal, get the baby reviewed by a more senior member of the team.
- If in doubt arrange an echocardiogram. (See internal echocardiogram guideline).

Key Points

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<tr>
<th>Key Points</th>
<th>Level of evidence</th>
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<tbody>
<tr>
<td>Routine newborn auscultation for murmurs is an insensitive method for early detection of congenital heart disease.</td>
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<tr>
<td>Routine pulse oximetry may improve the early diagnosis of some congenital heart lesions.</td>
<td>14,15</td>
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<tr>
<td>A low index of suspicion (based on all cardiac signs) and early referral for echocardiography are currently the best (and imperfect means) for early detection of congenital heart disease.</td>
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References


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