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Evaluation of suspected congenital heart disease in the neonatal period

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Summary Congenital heart disease (CHD) is the most common group of significant congenital abnormalities. It may present in the neonatal period with an asymptomatic murmur detected on the routine neonatal examination or when an infant becomes symptomatic. In assessing an infant with possible CHD, key features in history and examination need to be considered. The investigations appropriate to consider outside the tertiary paediatric cardiology setting will be discussed. We highlight the importance of a thorough assessment of the neonate presenting with an asymptomatic heart murmur to ensure that, where possible, infants with duct-dependent CHD are not discharged home inappropriately. The symptomatic presentation of CHD in the neonatal period is described, considering groups related by physiology rather than concentrating on the details of individual lesions. Important points in the initial stabilisation of a symptomatic infant are outlined.

Practice points

- Cyanosis is easily missed, always check oxygen saturation.
- Loud murmurs (especially associated with thrills) or abnormal pulses must always be taken seriously.
- Dysmorphic features or the presence of associated anomalies increase the risk of a murmur being pathological.
- Always consider congenital heart disease in the sick infant.

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Introduction

Congenital heart disease (CHD) can be defined as a structural abnormality of the heart or intrathoracic great vessels which is actually or potentially of functional significance.[1] It represents a spectrum of conditions, from those that may be fatal in the neonatal period, to those with which a normal lifespan would be expected.

Current guidelines recommend initial screening for CHD in the neonatal period with a further examination at 6–8 weeks.[2] Around 45% of CHD will be detected on routine neonatal examination when a large number of innocent murmurs will also be heard. Even severe lesions may not be detected at this point due to the presence of a widely patent ductus arteriosus.[3] Many lesions, particularly those

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with significant left ventricular outflow tract obstruction will become symptomatic before the second examination and carry a high mortality if they continue to go unrecognised.[4,5] The appropriate assessment of infants who have been found to have an abnormal neonatal cardiac examination and an awareness regarding the range of presentations of CHD in early infancy are therefore vital.

Risk factors for CHD

CHD is the most common group of significant congenital malformations accounting for 40% of the total group.[6] Most studies estimate the incidence of CHD to be between 5 and 8 per 1000 live births, although this figure varies according to case definition and method of case ascertainment.[7]

Family history

Having a sibling with CHD confers a 2% risk on a subsequent child. A mother with CHD has a 6% risk of having an affected offspring and a father a 2% risk overall.[8] There is some variation in these figures depending on the specific lesion present.

Syndromes and associations

The aetiology of CHD is complex, and in most cases multifactorial. There are a number of recognised associations that include:

- Chromosomal abnormalities (Down, Edward, Patau, Turner, cri-du-chat)
- Contiguous gene syndromes (William's, DiGeorge's)
- Single gene defects (Noonan's, Marfan's, isomerisms)
- Teratogens (anticonvulsants, alcohol, lithium)
- Congenital infection (rubella)

Even in conditions, such as Down syndrome, in which the association with CHD is clear, the mechanisms underlying the abnormality are only now beginning to be elucidated.

Antenatal detection

Routine antenatal screening for CHD is limited to a four-chamber view of the heart and is performed at around 20 weeks gestational age as part of anomaly scanning. This will pick up around 25% of significant CHD,[9] although with specific training and assessment of the ventricular outflow tracts, this may be significantly improved.[10] Fetal echocardiography, in the experienced hands, is both sensitive and specific however, its role is limited to those 5% of pregnancies in which an abnormality has been detected on routine antenatal scanning or in which an identified risk factor is present.[10,11] In the majority of cases with no previously identifiable risk factor CHD will continue to go undetected prior to delivery.

Presentation of CHD in the neonatal period

In the neonatal period a diagnosis of CHD may be considered for two reasons: (1) a heart murmur or other cardiovascular abnormality identified in an asymptomatic infant or (2) the development of symptoms and signs that could be attributable to CHD. The initial assessment of these infants will be discussed.

Assessment of a child with suspected CHD

History

The following information needs to be obtained at the initial review:

- *Risk factors*: Family history, maternal medication or congenital infection.
- *Antenatal scans*: Routine anomaly and fetal echocardiography.
- *Perinatal*: Considering risk factors for infection and persistent pulmonary hypertension that may present in a way indistinguishable from CHD.
- *Postnatal*: Breathing and feeding difficulties. Excess weight gain or failure to lose weight after birth.

Examination

A full examination should include:

- *Evaluation of airway, breathing and circulation*: Assessment and management of life-threatening problems.
- Presence of dysmorphic features and other congenital abnormalities.
- *Colour*: Cyanosed or pale. Check oxygen saturation with a pulse oximeter.