How effectively can clinical examination pick up congenital heart disease at birth?

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Perspective on the paper by Patton and Hey (see page 263)

Congenital heart disease (CHD) is a major cause of death in infancy in term babies. It accounts for 3% of all infant deaths and 46% of all deaths from congenital malformations.¹ Those surviving beyond infancy have a 96% chance of reaching 16 years.² It is for these reasons that screening for CHD is essential. Routine antenatal screening for congenital heart defects is performed as part of the general anomaly scan at 18–20 weeks gestation. At present, this identifies on average about 25% of affected fetuses. Because of this low yield, routine clinical examination of all newborn babies remains necessary with the expectation that those with heart defects will be picked up.

The vast majority of these early deaths, particularly those occurring in the first two weeks of life, are due to a handful of lesions. They are the so called duct dependent lesions, namely coarctation of the aorta, critical aortic stenosis, interrupted aortic arch, hypoplastic left heart syndrome, transposition of the great arteries, pulmonary atresia, and critical pulmonary stenosis. To these must be added obstructed total anomalous pulmonary venous connection. These lesions, although individually rare, form the bulk of the life threatening heart conditions in the newborn period. The aim of any screening examination should therefore be to identify them.

To date, the reported results of such examinations have been uniformly poor from various studies. In one study, routine neonatal examination failed to detect more than half of babies with heart defects of all types.³ In contrast, in this issue of the journal a study from Ashington in the North of England shows what can be achieved.⁴ This service was developed as a deliberate experiment after the loss of junior doctor cover. The paper reports the results of the first eight years of the service. It is a service delivered by nurse practitioners with maximum support and training from neonatal consultant paediatricians and the regional paediatric cardiac centre. It shows an initial learning curve followed

in the second half of the study period with quite impressive results. They achieved a sensitivity of 94.3% and a specificity of 97.7% in their referral process, and no babies with a life threatening condition were missed in the second half. Fewer complex and life threatening conditions were recorded, however, in the second half of the study period.

Why are the results of neonatal examinations poor, and what lessons can we learn from what has been described elsewhere as the Ashington experiment?⁵ Many factors account for this.

Symptoms and signs of heart failure are very uncommon in the immediate newborn period in babies with structural heart disease. Many newborn babies with severe heart disease can look quite well at birth. For this reason surrogate markers of heart disease, particularly those that suggest the aforementioned life threatening conditions, are usually looked for. These include cyanosis, murmurs, and poor pulses. Cyanosis can be quite difficult to recognise clinically except where it is profound. In babies with adequate pulmonary blood flow and saturations in the low 90s, it is often a challenge to make a diagnosis on clinical assessment alone. Murmurs, if present, may herald critical CHD, but they are sometimes completely absent. Lower limb pulses in babies with obstructive lesions of the left side of the heart can be normal if the duct is still widely open with good right to left shunt through it.

The timing of the newborn examination can also have a bearing on the situation. Assessment performed within six hours of birth in those who are booked for early discharge is more likely to miss duct dependent lesions because in many babies the duct may still be widely patent. Examinations performed on the second or third day of life stand a much better chance of detecting an abnormality.

Neonatal examinations are usually carried out by senior house officers. Sometimes these doctors are in the first six months of their paediatric training.

Most doctors can remember how daunted and vulnerable they felt in the first few months of their training when faced with a newborn baby. More recently in the United Kingdom, advanced neonatal nurse practitioners have been given responsibility in some units for neonatal examinations. The reasons for this trend are various. The change in junior doctors' hours has made it more difficult in some units to provide this service using medical personnel. In others the service has been devolved because the unit has lost accreditation as it is no longer able to sustain training posts.

Irrespective of who is delivering the service, there are important requirements. The first essential requirement is adequate training of the personnel to make sure that they know what to look for and what to do when it is found. One of the differences between nurse and doctor delivered services is that quite often doctors are simply told what to do and left to do it on the basis that their basic training has equipped them with the skills to adapt to the demands of the job. Nurses, in contrast, always go through a process of mentoring and evaluation on the job before they are left on their own. It is salutary to note that this approach is now being adopted in the training of junior doctors in the United Kingdom.

The second essential requirement is good communication between members of the obstetric, neonatal, and cardiac units involved in the service, as well as between them and the community primary care services. This will ensure that babies are seen promptly and referred appropriately if problems are identified. This does not always happen.³

The third, and perhaps the most important, requirement is parent education on the features of early heart disease in the newborn baby. This should be started in the antenatal period when parents are relaxed and can ask questions and give feedback to make sure that they have understood the education given. This can then be reinforced at the time of the neonatal examination before discharge. Screening examinations by their nature are not diagnostic tests.⁶ There will always be false positive and false negative findings. Education should highlight this and ensure that parents recognise the vital role that they have in making sure that those babies who will inevitably be missed do not come to harm.

Finally it needs to be pointed out that clinical examination on its own will never be entirely adequate. The ideal would be for all babies to have a screening echocardiogram before discharge. The resources for this would be

enormous and are unlikely to be achievable in the foreseeable future. There are now studies that show that pulse oximetry enhances and improves the results of clinical screening. This is the conclusion drawn by the recently published *Health Technology Assessment* report on newborn screening for congenital heart defects.⁶ It calls for studies to evaluate this combination as a cost-effective model for future screening. It is necessary to point out that comparisons between preductal and postductal saturations need to be made for maximum effectiveness.

What are the lessons from Ashington? Firstly, it underlines the utility of a clear protocol. Secondly, and without undervaluing the enormous support given by the community and management to the service,⁵ it shows the importance of

those delivering care having a sense of ownership of the service. Thirdly, in the wider context, it shows that a nurse delivered neonatal screening service can be effective and gives hope to those parts of the world where shortage of medical personnel could be deemed a problem. Previous studies have already shown that neonatal screening examinations performed by nurses are as good as those done by senior house officers.⁷

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