

Prenatal diagnosis of structural heart disease: does it make a difference to survival?

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Is there a survival advantage conferred by the prenatal diagnosis of congenital heart disease?

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There are two potential advantages of mid trimester diagnosis of fetal heart disease. Perinatal management in an environment where the appropriate expertise is available and prepared might result in improved outcomes after intervention in the newborn period in situations where this is required. Alternatively, prenatal diagnosis of congenital heart disease allows consideration of termination of pregnancy. Termination of pregnancy for fetal anomaly is an emotive subject and has a wide range of acceptability in different societal contexts. Nearly 20% of all pregnancies in the UK were terminated for social indications in 1994 whereas the proportion in which the indication for termination was detection of fetal cardiac abnormality was about 0.02%.¹

SURVIVAL ADVANTAGE?

If there is a survival advantage conferred by the prenatal diagnosis of congenital heart disease, its demonstration has been elusive. The spectrum of heart abnormalities diagnosed prenatally differs from that seen in postnatal practice. Complex cardiac abnormalities, associated extracardiac abnormalities, and chromosome defects are over represented.² It is not therefore surprising that outcomes for unselected series of structural cardiac abnormalities diagnosed prenatally have been poor, with high rates of spontaneous intrauterine or early neonatal death in continuing pregnancies.

Intuitively, it might be thought that the most likely survival advantage conferred by prenatal diagnosis would occur in isolated cardiac abnormalities in which the natural history would be for early neonatal death to occur because of dependency of either systemic or pulmonary blood flow on continued patency of the arterial duct. Early encouragement for this view was provided by data which indicated that neonates born with hypoplastic left heart syndrome after prenatal diagnosis had a lower incidence of severe preoperative acidosis before initial palliative surgery when compared to historic controls.³ However, the probability of postnatal survival for live born infants with pulmonary atresia and intact ventricular septum was similar regardless of the timing of diagnosis in a large population based study.⁴ The hypothesis that costs and duration of initial hospitalisation and survival to discharge home would be improved by prenatal

diagnosis was examined in a cohort analysis of neonates with structural heart disease in the absence of other life threatening conditions.⁵ There was no difference in survival among those requiring palliative surgery because of a functionally single ventricle. Moreover, the costs and duration of hospitalisation were greater in the group diagnosed prenatally, regardless of whether or not surgery was performed postnatally. The results were similar when the analysis was limited to infants whose survival was dependent on continued patency of the arterial duct.

HYPOPLASTIC LEFT HEART

The heart abnormality diagnosed prenatally more than any other is hypoplastic left heart.¹ Staged palliative surgery for this condition is available. Inevitably, parental decisions following prenatal diagnosis depend crucially on the counselling received regarding the likely postoperative outcome. The literature may be difficult to interpret. In a unit with large experience of this type of palliative surgery, early survival after prenatal diagnosis of hypoplastic left heart was worse than might have been predicted from the results of a contemporaneous surgical series from the same institution.^{6,7} This can be accounted for in part by spontaneous intrauterine death in continuing pregnancies and unsuitability for palliative surgery postnatally despite an intention for surgical treatment.⁸ In addition, infants born with undiagnosed structural heart abnormalities need to survive sufficiently long to reach an appropriate surgical centre in order to qualify for inclusion in a surgical series. Consequently, infants diagnosed postnatally who then undergo surgery, or assessment for surgery, are already a selected group in comparison to newborn infants in whom the same cardiac diagnosis was known prenatally. These factors may also explain why other studies of surgical patients have failed to demonstrate improved postoperative survival after prenatal diagnosis of hypoplastic left heart.⁹⁻¹¹ A recent report indicating improved hospital survival after initial staged palliative surgery for hypoplastic left heart following prenatal diagnosis¹² is more encouraging, but may not be directly comparable with previous data^{6,8} as infants with birth weight less than 2 kg or serious extracardiac abnormality were excluded. In a report from a single surgical institution it is also difficult to exclude a selection bias in referral of prenatally diagnosed cases of hypoplastic left heart perceived to be good risk surgical candidates. Population based data may be better able to address this question.¹

None of the studies cited above was designed to detect mortality resulting from neonatal death

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with congenital heart disease not diagnosed during life. Detection of this occult cardiac mortality allowed demonstration of improved survival after prenatal diagnosis of transposition of the great arteries in Paris.¹³ By analogy, four critically ill newborn infants with respiratory failure and undiagnosed transposition of the great arteries have been referred to Great Ormond Street Hospital for ECMO (extracorporeal membrane oxygenation) support since 1994. However, the impact of prenatal diagnosis on improved postnatal survival will not be great in population terms while as little as 3% of all cases of transposition of the great arteries with intact ventricular septum are diagnosed prenatally.¹ The study from Oxford reported in a recent issue of *Heart*¹⁴ similarly sought to detect deaths in infants with previously undiagnosed heart disease in an attempt to demonstrate a survival advantage after prenatal diagnosis of coarctation of the aorta. The analysis was hampered by the small numbers, but the data suggest that survival was improved. There was an impressively low false positive diagnosis rate and a high detection rate, confirming the feasibility of prenatal detection of coarctation which often has quite subtle abnormalities on prenatal ultrasound examination. Avoidance of severe preoperative acidosis, which was afforded by prenatal diagnosis of coarctation in this group, may have minimised the severity of potential late neurodevelopmental morbidity¹¹ but this remains to be demonstrated.

PRESENT STATUS

Examination of the fetal heart has become an established component of mid trimester anomaly scanning, although specific practice varies in different healthcare systems.¹⁵ There is discussion whether screening recommendations should extend beyond a single four chamber view to also include assessment of the ventricular outflow tracts of the fetal heart. Skilled obstetric ultrasonographers will doubtless extend their examination of the heart beyond the four chamber view. However, there are important teaching, training, and audit resource issues if wider examination of the fetal heart becomes a general requirement. Serious structural heart disease might be expected to occur in only about two of every 1000 pregnancies.¹ Experience in the detection of fetal structural heart disease of an individual obstetric ultrasonographer will necessarily therefore be limited. An alternative strategy which may provide a higher yield in terms of prenatal diagnosis of structural heart abnormalities is first trimester nuchal translucency screening. If initial data are confirmed, structural heart disease may be detected by detailed fetal echocardiography in about 6% of fetuses in whom first trimester nuchal translucency has exceeded the upper 99% confidence interval and in whom aneuploidy has been excluded.¹⁶ The mechanistic association between increased nuchal translucency and structural heart disease is not known.

It remains difficult to advocate a population screening programme for prenatal diagnosis of structural heart disease on the basis that this will result in improved outcome after postnatal intervention. Nevertheless, more than 50% of all cases of

hypoplastic left heart and univentricular atrioventricular connection in the population are now diagnosed prenatally in the UK.¹ This allows considered decision making after detailed fetal echocardiography, and potential self selection of families best able to cope with the rigours of staged palliative surgery, or even possible prenatal listing for transplantation. Procedure related mortality has decreased steadily for most neonatal and infant cardiac operations, and studies addressing late morbidity and quality of life issues are now a priority, partly to aid counselling after prenatal diagnosis in this situation. For the presently small but probably increasing population with prenatal diagnosis of transposition of the great arteries or coarctation of the aorta, conditions which pose a risk of neonatal death but which are amenable to biventricular repair, the potential long term dividend is even greater.

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