Newborn screening for congenital heart defects

Newborn screening for congenital heart defects: a systematic review and cost-effectiveness analysis

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Executive summary

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Executive summary

Objectives

The objectives of this study were to provide evidence to inform policy decisions about the most appropriate newborn screening strategy for congenital heart defects and to identify priorities for future research that might reduce important uncertainties in the evidence base for such decisions.

Specifically the study aimed to:

- systematically review the epidemiology, natural history, treatment and outcomes of congenital heart defects, as well as the performance, effects and costs of current and alternative newborn screening strategies
- classify congenital heart defects for newborn screening taking into account clinical features, presymptomatic interval, prevalence, natural history and treatment
- evaluate effects, costs and cost-effectiveness of alternative newborn screening strategies
- explore the values of parents and health professionals towards the quality of life of children with congenital heart defects
- explore parental experiences of newborn screening for, and diagnosis of, congenital heart defects.

Methods

A systematic review of the published medical literature concerning outcomes for children with congenital heart defects was carried out. The results of this review were then used in the decision analytic model, based on a population of 100,000 live-born infants, developed to assess the cost-effectiveness of alternative screening strategies for congenital heart defects relevant to the UK.

A study was then carried out exploring the perspectives of parents and health professionals towards the quality of life of children with congenital heart defects. Eight health state descriptions of degrees of cardiac and neurological disability resulting from congenital heart defects were developed and these were presented with a self-administered anonymous questionnaire to two groups of respondents: parents of a child with a congenital heart defect and the health professionals who care for them. Respondents were asked to rank and then score these health states on a visual analogue scale; they then marked the state 'death' on the scale. The views of health professionals and parents about the quality of life of children with congenital heart defects, as represented by these typical health states, were compared.

Finally, a structured review was carried out of the medical literature regarding parental experiences of newborn screening with relevance to screening for congenital heart defects. The findings from the literature review were linked with those from a focus group set up by the study with parents of children with congenital heart defects.

Results

Epidemiology

Congenital heart defects affect 7–8 per 1000 live-born infants and account for 3% of all infant deaths and 46% of deaths due to congenital malformations. Around 18–25% of affected infants die in the first year, with 4% of those surviving infancy dying by 16 years.

Outcomes

Long-term sequelae include cardiac arrhythmias, infective endocarditis and pulmonary vascular obstructive disease.

The study found that long-term outcome studies addressing physical disability, neurodevelopmental, cognitive or psychosocial outcomes and the capacity to participate in normal childhood activities are lacking. Severe neurological deficits affect 5–10% following surgery and milder neurological problems occur in up to one-quarter of children.

Classification of congenital heart defects

Congenital heart defects can be classified into three main types. ▶

- Life-threatening congenital heart defects are structural cardiac malformations in which collapse is likely and comprise: transposition of the great arteries, coarctation/interrupted aortic arch, aortic stenosis, pulmonary atresia and hypoplastic left heart/mitral atresia.
- Clinically significant congenital heart defects are structural cardiac malformations that have effects on heart function but where collapse is unlikely or its prevention unlikely to be feasible. The most common defects in this group are ventricular septal defect, complete atrioventricular septal defect, atrial septal defect and tetralogy of Fallot.
- Clinically non-significant congenital heart defects are anatomically defined cardiac malformations that have no functional clinical significance. They include ventricular septal defects only detectable with echocardiography and requiring no treatment.

Screening

The primary objective of newborn screening is the presymptomatic identification of life-threatening congenital heart defects to achieve a timely diagnosis, defined as a preoperative diagnosis before collapse or death occurs. A secondary objective is the detection of clinically significant congenital heart defects.

Current newborn screening policy comprises a clinical examination at birth and 6 weeks, with specific cardiac investigations for specified high-risk children. Routine data are lacking, but under half of affected babies, not previously identified antenatally or because of symptoms, are identified by current newborn screening. There is evidence that screen-positive infants do not receive timely management.

Pulse oximetry and echocardiography, in addition to clinical examination, are alternative newborn screening strategies but their cost-effectiveness has not been adequately evaluated in a UK setting.

Decision analysis

In a population of 100,000 live-born infants, the model predicts:

- 121 infants with life-threatening congenital heart defects undiagnosed at screening, of whom 82 (68%) and 83 (69%) are detected by pulse oximetry and screening echocardiography, respectively, but only 39 (32%) by clinical examination alone. Of these, 71, 71 and 34, respectively, receive a timely diagnosis
- 46 (0.5%) false-positive screening diagnoses per 100,000 infants with clinical examination, 1168 (1.3%) with pulse oximetry and 4857 (5.4%) with screening echocardiography. The latter includes infants with clinically non-significant defects
- total programme costs of £300,000 for clinical examination, £480,000 for pulse oximetry and £3.54 million for screening echocardiography.

The additional cost per additional timely diagnosis of life-threatening congenital heart defects ranges from £4900 for pulse oximetry to £4.5 million for screening echocardiography. Including clinically significant congenital heart defects gives an additional cost per additional diagnosis of £1500 for pulse oximetry and £36,000 for screening echocardiography. Key determinants for cost-effectiveness are detection rates for pulse oximetry and screening echocardiography.

Valuing quality of life

Parents and health professionals place similar values on the quality of life outcomes of children with congenital heart defects and both are more averse to neurological than to cardiac disability.

Parental views

Adverse psychosocial effects for parents are focused around poor management and/or false test results.

Conclusions

The main conclusions of the study are as follows.

- Early detection through newborn screening potentially can improve the outcome of congenital heart defects.
- The current programme performs poorly, and lacks monitoring of quality assurance, performance management and longer term outcomes.
- Pulse oximetry is a promising alternative newborn screening strategy but further evaluation is needed to obtain more precise estimates of test performance and to inform optimal timing, diagnostic and management strategies.
- Although screening echocardiography is associated with the highest detection rate, it is the most costly strategy and has a 5% false-positive rate.
- Improving antenatal detection of congenital heart defects increases the cost per timely postnatal diagnosis afforded by any newborn screening strategy but does not alter the relative effects of the strategies.
- Timely management of screen-positive infants is essential if outcomes are to improve. ▶

Implications for health care

The findings suggest the following:

- Broadly, newborn screening for congenital heart defects meets the National Screening Committee criteria for a screening programme.
- There is a strong case for modifying the current policy of clinical screening of the newborn and 6-week-old infant to include other more effective tests.
- The review and the decision analysis suggest that pulse oximetry in addition to clinical examination appears to be a strong candidate for screening, but would require further research evaluation to inform policy.
- Adequate diagnostic and management services are essential to ensure good outcome.
- Information for parents and health professionals is needed across the antenatal and newborn continuum, as is a training curriculum for midwives and others involved in screening.
- Routine data systems, currently lacking, are required for audit, quality assurance and to assess longer term follow-up, as are clearly defined process and outcome measures.

Recommendations for further research

The following areas are suggested for further study:

- Refining the detection rate and other aspects of pulse oximetry.
- More direct evaluation of antenatal screening strategies.
- Investigating the psychosocial effects of newborn screening for congenital heart defects.

Publication

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Research suggestions are carefully considered by panels of independent experts (including service users) whose advice results in a ranked list of recommended research priorities. The HTA Programme then commissions the research team best suited to undertake the work, in the manner most appropriate to find the relevant answers. Some projects may take only months, others need several years to answer the research questions adequately. They may involve synthesising existing evidence or conducting a trial to produce new evidence where none currently exists.

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The research reported in this monograph was commissioned by the HTA Programme as project number 99/45/01. The contractual start date was in March 2001. The draft report began editorial review in February 2004 and was accepted for publication in February 2005. As the funder, by devising a commissioning brief, the HTA Programme specified the research question and study design. The authors have been wholly responsible for all data collection, analysis and interpretation, and for writing up their work. The HTA editors and publisher have tried to ensure the accuracy of the authors' report and would like to thank the referees for their constructive comments on the draft document. However, they do not accept liability for damages or losses arising from material published in this report.

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