Economic evaluation of neonatal screening for phenylketonuria and congenital hypothyroidism

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Record Status
This is a critical abstract of an economic evaluation that meets the criteria for inclusion on NHS EED. Each abstract contains a brief summary of the methods, the results and conclusions followed by a detailed critical assessment on the reliability of the study and the conclusions drawn.

Health technology
The authors assessed a routine neonatal screening programme and compared this with symptomatic diagnosis of phenylketonuria (PKU) and congenital hypothyroidism (CH). The screening programme comprised a heel prick to newborns.

Type of intervention
Screening.

Economic study type
Cost-effectiveness analysis.

Study population
The study population comprised newborns. No specific inclusion or exclusion criteria were reported.

Setting
The setting was the community. The economic study was carried out in Western Australia.

Dates to which data relate
The authors used epidemiological data, which did not appear to relate to a specific year, and other effectiveness data that were derived from studies published between 1984 and 2003. The dates when some of the resource use data were collected were not reported; other data were taken from studies published between 1963 and 2003. The price year was 2001.

Source of effectiveness data
The effectiveness data were derived from completed studies.

Modelling
A mathematical model was developed to estimate the cost per annum for intellectual disability. This cost was related to the proportion of individuals living at home, the cost of early intervention, the cost of annual respite, the cost of a leisure buddy programme, the cost of an alternative to employment, and the cost of out-of-home care.

Outcomes assessed in the review
The main outcomes estimated from the review of the literature were the levels of intellectual disability for cases of PKU and CH undetected at birth.
Study designs and other criteria for inclusion in the review
Not reported.

Sources searched to identify primary studies
Not reported.

Criteria used to ensure the validity of primary studies
Not reported.

Methods used to judge relevance and validity, and for extracting data
Not reported.

Number of primary studies included
Two studies were included in the review.

Methods of combining primary studies
The primary studies were not combined.

Investigation of differences between primary studies
No differences between the primary studies were investigated.

Results of the review
Levels of intellectual disability for cases of PKU undetected at birth were 64% severe and 36% moderate. The proportions for CH were 15% severe, 25% moderate, 40% mild or borderline, and 20% normal.

Methods used to derive estimates of effectiveness
The authors used epidemiological data to estimate the effectiveness of screening. They also made an assumption, based on a single published article, to inform life expectancy.

Estimates of effectiveness and key assumptions
The authors stated that, based on the expected number of cases each year, an average of 9.5 children are born either with PKU (1.8 children) or CH (7.5 children) each year.

There was an underlying assumption that screening would identify all of these children, although this was not explicitly stated.

Life expectancy was assumed to be 74.0 years for mild levels of intellectual disability, 67.6 years for moderate levels of intellectual disability, and 58.6 years for severe levels of intellectual disability.

Measure of benefits used in the economic analysis
The authors had planned to estimate changes in life-years and changes in quality of life. However, as the screening programme led to net savings in costs, they determined that estimating a summary measure of health for the purpose of a cost-effectiveness ratio was not useful.
**Direct costs**

The authors reported that they analysed costs from the perspective of the public sector. From the unit costs estimated, this seems to have encompassed primarily the costs to the health care provider. The costs estimated were:

- programme costs including specimen collection costs (i.e. nursing and consumables) and sample testing (i.e. equipment costs, overheads, salaries and consumables, and building costs);
- costs of intellectual disability based on expected levels of intellectual disability for PKU and CH; and
- costs of maternal PKU, including tracking and educational costs and pregnancy costs.

The treatment costs were assumed to be the same for early (screening) and late detection (symptomatic diagnosis) and so were not included in the analysis. No additional treatment costs for residual effects of treated conditions were included, as residual effects (developmental delay and behavioural problems) were reported to be within the normal developmental range. The costs of the programme were based on the Western Australian screening programme and 25,000 screened babies each year. The cost of intellectual disability was derived from a review of the literature (studies published between 1963 and 2003), which provided inputs to a mathematical model. The cost of maternal PKU was based on information provided from institutions and cases reported in the study setting. All costs were reflated to 2001 prices using health index deflators. The costs incurred in future years were discounted at a rate of 5%.

**Statistical analysis of costs**

The costs were treated deterministically.

**Indirect Costs**

The authors acknowledged that indirect costs, in terms of productivity lost due to illness, were relevant to the analysis but noted that the analysis was limited to the direct costs. This was in keeping with the perspective adopted for the analysis.

**Currency**

Australian dollars (AUD).

**Sensitivity analysis**

A one-way sensitivity analysis was used to explore the impact of different levels of intellectual disability associated with children with PKU and CH, as well as the discount rate used to value future costs.

**Estimated benefits used in the economic analysis**

See the 'Effectiveness Results' section.

**Cost results**

The total discounted programme cost for 25,000 babies screened annually was AUD 569,672 (at an annual discount rate of 5%).

The total discounted cost averted due to avoiding PKU and CH in 1 year was AUD 3,449,448.

The net annual cost-savings were AUD 2,879,776 (at an annual discount rate of 5%).

**Synthesis of costs and benefits**

Not relevant.
Authors' conclusions
The authors concluded "this analysis has shown that newborn screening for PKU (phenylketonuria) and CH (congenital hypothyroidism) provides a net gain in terms of benefits to individuals and their families and a financial benefit to government services and hence the community as taxpayers".

CRD COMMENTARY - Selection of comparators
The authors compared routine neonatal screening with symptomatic diagnosis. They acknowledged the difficulties in making such comparisons due to the fact that the universal screening programme already in place, but there was an implicit assumption underlying the analysis that screening would identify all potential cases.

Validity of estimate of measure of effectiveness
The authors used epidemiological data, data from published studies, and assumptions to estimate effectiveness. The assumptions seem to have been based on the best information available and were tested in sensitivity analyses.

Validity of estimate of measure of benefit
The authors did not estimate a summary measure of benefit. The study was, in effect, categorised as a cost-consequences analysis.

Validity of estimate of costs
A public sector perspective was adopted for the cost analysis and the estimates carried out reflected that perspective. The authors estimated the total costs associated with the screening programme that would impact on public sector funding, and compared this with the estimated cost of treating individuals with PKU and CH. The authors identified very large cost-savings associated with the screening programme. Although there was no statistical analysis to determine whether the differences were statistically different, the large physical difference suggests that small omissions in costs may not have affected the principal results and thus conclusions. The analysis was well reported with a breakdown of the unit costs and a clear statement that the costing was based on the screening of 25,000 babies. The authors reported discounted and undiscounted costs for the treatment of PKU and CH, enabling greater transparency of their results.

Other issues
The authors were able to compare their own results with findings from other studies, suggesting that this evaluation supported previous analyses. The issue of generalisability to other settings was not addressed. Since the authors used costs and practices local to Western Australia, generalisability may be limited to this area, although the use of sensitivity analyses improves the generalisability of the results. Readers might consider their own setting and assess the comparability to establish whether there is the possibility to extrapolate the results. Part of the objective of the study was to ensure that the "benefits accrued outweigh the associated burden to the larger population". In order to assess this objective more fully, the authors might have considered the decrement in quality of life for the babies and their parents associated with screening for everyone, and compared this to the increase in quality of life for the small number for whom PKU and CH are diagnosed before they become symptomatic. However, this would have required a societal perspective be adopted, and so the public sector perspective that was actually adopted was insufficient to address the study question.

Limitations focused on the inability to prospectively compare unscreened cases, as it is no longer ethical to randomly not screen. The authors' conclusions, that they were able to show that screening provided a net gain to individuals and their families, was not strictly true as the authors did not set out to achieve this and primarily used assumptions to demonstrate the benefits of screening. These benefits were not estimated within this study.

Implications of the study
The authors remarked that ongoing evaluation is necessary to ensure the optimum use of resources. However, it would appear that this work should focus on different methods of achieving screening and their associated costs, rather than a...
cost-effectiveness analysis of screening compared with no screening.

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