Expanded newborn screening for genetic and metabolic disorders: modeling costs and outcomes
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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.

CRD summary
The objective was to examine the clinical and economic impact of adding to the current screening programme for newborns, two genetic or metabolic tests for medium-chain acyl-coenzyme A dehydrogenase deficiency and β-ketothiolase deficiency. The authors concluded that their findings might help decision makers to evaluate the addition of screening to the current newborn programme. The study was not extensively reported and had some methodological limitations, which may affect the validity of the authors’ conclusions.

Type of economic evaluation
Cost-effectiveness analysis

Study objective
The objective was to examine the clinical and economic impact of adding to the current screening programme for newborns, a screening for two genetic or metabolic disorders, which were medium-chain acyl-coenzyme A dehydrogenase deficiency (MCADD) and β-ketothiolase deficiency (BKT).

Interventions
The two additional screening tests were compared against no additional screening. The screening procedure was based on tandem mass spectrometry. Those who tested positive received a confirmation test, which was assumed to be the gold standard.

Location/setting
USA/hospital.

Methods
Analytical approach:
This economic evaluation was based on a decision analytic model. The time horizon of the analysis was short and corresponded to the screening test turn-around time. The authors did not state explicitly the perspective that was adopted.

Effectiveness data:
The clinical data came from a selection of known, relevant studies and the Maryland 2005 newborn screening programme, which provided testing and follow-up data for approximately 70,000 babies. In general, US studies were selected for the clinical and epidemiological estimates. Expert opinions were also used when published data were not available. The key clinical endpoint was the accuracy of screening (its sensitivity and specificity).

Monetary benefit and utility valuations:
Not relevant.

Measure of benefit:
No summary benefit measure was used as a cost-consequences analysis was carried out. The main clinical outputs of the model were the rates of true- and false-positives and true- and false-negatives for each screening test.

Cost data:
The economic analysis included the costs of screening such as the instruments, labour, operational expenses, and confirmatory test (including time costs for parents). The cost of a false-positive result of screening included the immediate response by a nurse coordinator, a visit to an urgent care centre, laboratory tests, and consultation with a geneticist. The costs and quantities of resources used were derived from the literature as well as from expert opinion and personal communications. They were in US dollars ($) and the price year was 2005.

Analysis of uncertainty:
A deterministic univariate sensitivity analysis investigated the impact of changes in the model inputs, such as the prevalence of disease and accuracy of screening.

Results
With a prevalence of 1:17,000, in a cohort of 100,000 newborns, MCADD screening would result in 5.9 true-positives, 139.91 false-negatives, 99,854 true-negatives, and 0 false-negatives. The cost per newborn would be $22.77.

With a prevalence of 1:100,000, in a cohort of 100,000 newborns, BKT screening would result in 1 true-positive, 289.6 false-negatives, 99,709 true-negatives, and 0 false-negatives. The cost per newborn would be $3.08.

Thus, in Maryland in a cohort of 70,000 newborns, the expected number of cases of MCADD detected with the screening test (compared with no screening) was approximately 4, the expected number of false-positives was 98, and the total cost was $1,593,000. Similarly, the expected number of cases of BKT detected with the screening test (compared with no screening) was approximately 1, the expected number of false-positives was 204, and the total cost was $215,600.

The sensitivity analysis revealed that the specificity of screening was the most influential model input.

Authors' conclusions
The authors concluded that their findings might help decision makers to evaluate the addition of MCADD and BKT screening to the current newborn screening programme.

CRD commentary
Interventions:
The current newborn screening programme was implicitly considered as the background comparator. The costs and clinical consequences of adding the two tests were considered as incremental to the current strategy. However, the authors noted that the content of the screening programme varied across the USA as each state had its own programme. The two diseases studied were ranked first (MCADD) and last (BKT), among 29 genetic or metabolic disorders, in a recent recommendation by the American College of Medical Genetics.

Effectiveness/benefits:
The clinical data came from several published studies. Except for the Maryland database, the authors did not provide any information on the design and other characteristics of the sources nor on the approach used to determine the estimates from among those reported in the literature. This means that it is not possible to judge the validity of the clinical data. Also, some of the key clinical inputs were based on expert opinions. No summary benefit measure was used.

Costs:
The perspective was not explicitly stated. The sources of costs were not clearly described. The unit costs were presented for some items, while other costs were reported as macro-categories. The price year was reported. Statistical analyses of costs were not carried out and only a few estimates were varied in the sensitivity analysis.

Analysis and results:
A synthesis of the costs and benefits was not required, given the cost-consequences framework. The authors noted that the main limitation of their analysis was the need for expert opinions, due to the scarcity of published evidence. This introduced some uncertainty into the decision analysis. Other potential drawbacks were the short-term horizon and the use of a cost-consequences analysis, which precluded the use of an explicit decision criterion. The sensitivity analysis
investigated only some aspects of uncertainty.

Concluding remarks:
The study was not extensively reported and had some methodological limitations, which may affect the validity of the authors' conclusions.

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