Title **Clinical Effectiveness and Cost Effectiveness of Neonatal Screening for Inborn Errors of Metabolism Using Tandem Mass Spectrometry: A Systematic Review**

Agency **NCCHTA, National Coordinating Centre for Health Technology Assessment**
Mailpoint 728, Boldrewood, University of Southampton, Southampton SO16 7PX, United Kingdom;
Tel: +44 2380 595586, Fax: +44 2380 595639

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Aim

To evaluate the clinical and cost effectiveness of tandem mass spectrometry (MS) based neonatal screening for inborn errors of metabolism (IEM).

Conclusions and results

Evidence from the reviews of IEM found that the UK screening program for phenylketonuria (PKU) was well established, and there was universal agreement that neonatal screening for PKU was justified. Of the many other disorders that can be detected by tandem MS, the best candidate condition for a new screening program, was medium-chain acyl-coenzyme A dehydrogenase (MCAD) deficiency. For many other IEM that can be detected by tandem MS, robust clinical evidence was limited. Cost-effectiveness analysis using economic modeling indicated that substituting the use of tandem MS for existing technologies for screening of PKU alone could not be justified. However, results from the economic modeling indicate that the addition of screening for MCAD deficiency as part of a neonatal screening program for PKU using tandem MS would be economically attractive. Using an operational range of 50,000 to 60,000 specimens per system per year, the mean incremental cost for PKU and MCAD deficiency screening combined using tandem MS from the model was – £23,312 for each cohort of 100,000 neonates screened. This cost saving is associated with a mean incremental gain of 59 life-years. Additional economic modeling using the available evidence does not support including other inherited metabolic diseases in a neonatal screening program.

Recommendations

The evidence appears to support the introduction of tandem MS into a UK neonatal screening program for PKU and MCAD deficiency combined. Tandem MS has the potential for simultaneous multidisease screening using a single analytical technique. Although the marginal cost of extending the program to include other conditions may be relatively small, the application of this new technology to PKU and MCAD deficiency screening does not imply the wholesale inclusion of all disorders detectable by tandem MS.

Methods

This review updates of two previous health technology assessment reports of neonatal screening for IEM. These reports have been updated by a systematic review of published research (between 1995 and January 2002) on neonatal screening of inherited metabolic disorders using tandem MS. This was supplemented by a search for economic literature and the application of a modeling exercise to investigate the economics of using tandem MS in a neonatal screening program in the UK.

Further research/reviews required

Further research should focus primarily on the long-term effectiveness of treatment strategies on adverse outcomes (disabilities and impairments) under conventional management and the potential impact of early diagnosis using tandem MS.

Written by Abdullah Pandor, ScHARR Technology Assessment Group, Sheffield, UK