



Title	Newborn Screening for Medium Chain Acyl-CoA Dehydrogenase Deficiency Using Tandem Mass Spectrometry: Clinical and Cost Effectiveness
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Aim

To conduct a systematic review of the clinical and economic literature on the use of tandem mass spectrometry (MS/MS) in newborn screening for medium chain acyl-CoA dehydrogenase deficiency (MCADD) as opposed to clinical diagnosis.

Conclusions and results

Twenty-one studies (15 full-length reports, 6 abstracts) of limited quality met the selection criteria for clinical evaluation. The clinical sensitivity and specificity of MS/MS-based screening for MCADD were very high. Most patients detected by newborn screening were asymptomatic, while most who presented clinically had potentially irreversible damage. The percentage of fatal cases among those diagnosed clinically was significantly higher than that among those diagnosed by screening. The economic review and analysis showed that screening results in more quality-adjusted life-years (QALY) and lower morbidity and mortality compared with no screening. Also, screening is cost effective compared to no screening, if willingness to pay is \$50 000 per QALY. The primary economic analysis using Canadian data showed that screening is cost effective if willingness to pay is \$20 000 per QALY. Not all parents may want their newborns screened for MCADD, but they should be informed about the risks and given the option to refuse.

Recommendations

Not applicable.

Methods

A systematic review of the clinical and economic literature was performed. For the clinical review, published and unpublished sources were searched for studies reported between 1995 and 2005. Two independent reviews selected cohort studies related to newborn screening for MCADD using MS/MS and studies comparing outcomes of MS/MS-based screening and clinical diagnosis. Information on incidence, clinical validity, and genetics of MCADD detected by MS/MS and clinical

diagnosis and the outcomes of disability and mortality were recorded. Study quality was assessed using QUADAS. The economic search was conducted from 1995 onwards. Two independent researchers selected studies and used the British Medical Journal (BMJ) 35-item checklist to assess quality. Data were extracted using a structured form.

Further research/reviews required

Further studies are needed on the long-term health consequences of MCADD, health improvements due to early detection, and health-related quality of life among MCADD patients.