

Title:	Urinary metabolic profiling for detection of metabolic disorders
Agency:	Medical Services Advisory Committee (MSAC) MDP 106 Commonwealth Department of Health and Ageing GPO Box 9849 Canberra ACT 2601 http://www.msac.gov.au
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Aim

To evaluate the safety, effectiveness and cost-effectiveness of urinary metabolic profiling (UMP) for the detection of metabolic disorders.

Results and conclusions

Safety

UMP is a non-invasive procedure that is not considered to impose safety issues for patients.

Effectiveness

Available evidence supported use of UMP to diagnose medium-chain acyl-CoA dehydrogenase deficiency (MCADD) in screened newborns and for diagnosis of glutaric aciduria type 1 (GA1) in symptomatic patients. This evidence was limited because investigations were not designed as diagnostic test studies or direct comparisons with other tests. UMP was shown to be a reliable diagnostic procedure in evidence from studies where comparisons were made between participants with previously diagnosed metabolic disorders and those who were disease-free. This evidence was limited by study design, and overall, quality was poor. Results from a quality assurance program showed that UMP was able to distinguish between urine samples from patients with known metabolic disorders and control samples. Results were limited because the analysis excluded determination of UMP performance among patients with suspected metabolic disorders.

Cost-effectiveness

Evidence was insufficient to assess the cost-effectiveness of UMP. The annual number of UMP procedures was forecast to be approximately 11,150 with a cost of \$1.1 to \$2.1 m.

Advice

MSAC has considered the strength of the evidence in relation to the safety, effectiveness and cost-effectiveness of urinary metabolic profiling compared with alternative investigations for the detection of metabolic disorders for the three following indications:

1. Asymptomatic newborns with a positive screening result suggestive of metabolic disorder.
2. Individuals with a clinical presentation suggestive of a metabolic disorder.

3. At-risk family members of patients with specific genetic metabolic disorder.

The evidence was limited by three factors:

- the rarity of the various metabolic disorders such that conventional high quality comparative diagnostic test studies are not possible
- the large number of metabolic disorders that can be diagnosed by urinary metabolic profiling
- that urinary metabolic profiling has been standard practice in Australia for many years.

MSAC finds that urinary metabolic profiling is either as safe or safer than alternative investigations to diagnose metabolic disorders.

MSAC finds that urinary metabolic profiling is effective in diagnosing metabolic disorders and is likely to be more effective than alternative investigations in allowing a timely diagnosis, especially in patients with undifferentiated presentations where a metabolic disorder is suspected or needs to be excluded.

Given the lack of high quality comparative evidence of effectiveness, a cost-effectiveness analysis could not be conducted and a budget impact analysis was performed. Whilst it is likely that downstream costs incurred by alternative investigations or delayed diagnosis are avoided by the use of urinary metabolic profiling, these could not be formally costed. MSAC considers it is likely that urinary metabolic profiling in carefully selected patients is cost-effective.

The current funding arrangements adequately capture the target population. MSAC advises that current public funding arrangements within the health care system should continue to be supported for this procedure.

—The Minister for Health and Ageing noted this advice on 8 December 2008—

Methods

MSAC conducted a systematic literature review pertaining to UMP for the detection of metabolic disorders.