INAHTA Briefs

Issue 2007/65

*Title* **Tandem Mass Spectrometry and Neonatal Blood Screening in Quebec: Summary Report**

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*Reference* 07-03. ISBN 978-2-550-48905-4 (print, French), 978-2-550-49399-0 (English summary).
www.aetmis.gouv.qc.ca/site/en_publications_2007.phtml

## Aim

To evaluate whether it would be pertinent to use tandem mass spectrometry (MS/MS) for neonatal blood screening in Quebec and to: 1) examine the relevance of replacing current screening methods for phenylketonuria (PKU) and tyrosinemia type 1 (TT1) by MS/MS and introducing neonatal screening for medium-chain acyl-CoA dehydrogenase deficiency (MCADD); and 2) analyze the main ethical, social, economic, and organizational issues.

## Conclusions and results

This report was prepared at the request of the Quebec Health and Social Services Ministry in the context of scientific debates and pressure in favor of adopting MS/MS for neonatal blood screening of inborn errors of metabolism. The review confirms the importance of a case-by-case analysis for each inborn error of metabolism. Available options depend on the specific characteristics and state of knowledge for each disease, and the applicability of the technological developments to these diseases. Even though there are gaps in the data, evidence supports the clinical utility of neonatal screening for the 3 diseases in question. As for the appropriateness of implementing MS/MS-based screening in Quebec, the situation differs according to the disease. For MCADD, MS/MS is the only technology available for neonatal screening, and its performance is one of the best for this particular condition. For PKU, the literature suggests that MS/MS yields fewer false positives than the current technology, but compared to the results observed in Quebec, this advantage would not be substantial. However, if MS/MS were used for MCADD screening, the technology transfer for PKU would avoid a duplication of analytical steps and would be efficient, according to the health economics literature examined. For TT1, MS/MS-based neonatal screening relying on both tyrosine and succinylacetone assays seems promising, but needs further validation. Furthermore, the judiciousness of a technology transfer and its optimal timing depend on several ethical, social, legal, economic, and organizational issues in addition to the scientific and technical considerations.

## Recommendations

Three separate scenarios are proposed for consideration by policy-makers: 1) conducting a pilot study; 2) postponing the introduction of MS/MS until after the necessary validation studies for TT1 screening have been completed; and 3) introducing MS/MS for PKU and MCADD screening, while, either undertaking gradual technology replacement for TT1, or maintaining the current methods until the results of the validation studies are available. Whichever option is chosen, MS/MS must not be implemented hastily, since other issues (ethical, economic, and organizational) first need to be resolved.

## Methods

Critical review of the scientific literature, of epidemiological data in Quebec, and of the grey literature; cost analyses; analysis of some ethical, social, legal, economic, and organizational issues.

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ISSN 1654-501X