

Health

Cadham Provincial Laboratory
Provincial Programs and Services

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www.gov.mb.ca/health/publichealth/cpl

September 22, 2011

Dear Practitioner:

Re: Expanded Newborn Screening

Starting September 26, 2011, expanded screening will be added to the newborn screening panel. Expanded screening employs tandem-mass spectrometry (MS-MS) to simultaneously detect dozens of different markers of potential metabolic disease in a baby's bloodspot. This expands the number of conditions that can be detected to include approximately 40 additional metabolic disorders. Like many other screening tests, a positive result on screening does not mean a metabolic condition exists, only that further assessment and perhaps testing is recommended. Some additional disorders that will be detectable include:

- Tyrosinemia
- Homocystinuria
- Citrullinemia
- Argininosuccinic acidemia
- Maple Syrup Urine Disease
- Medium chain acyl-CoA dehydrogenase (MCAD) deficiency, and
- Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency, among many others.
- Isovaleric acidemia
- Multiple Carboxylase Deficiency
- Glutaric Acidemia type 1
- Methylmalonic acidemia
- Carnitine Uptake Deficiency

Babies with suspicious results on expanded screening will generally be asked to provide a second bloodspot. The process is not different from the current second-request process. Babies with critically abnormal results will be immediately referred to Metabolic Diseases specialists at Winnipeg Children's Hospital for further assessment. The follow-up practitioner information at the bottom of the newborn screening card is critical for notifying caregivers of abnormal or critical results. Please ensure this information is provided on all newborn specimens.

Screening for galactosemia, biotinidase deficiency, congenital hypothyroidism, congenital adrenal hyperplasia, and cystic fibrosis will continue. Screening for PKU will be conducted using MS-MS and, instead of having a separate result, will be reported with the expanded screening results. Updated newborn screening English/French pamphlets for parents are now available from Materials Distribution Agency or CPL shipping. Further information is available effective September 26, 2011, at: <http://www.gov.mb.ca/health/publichealth/cpl/index.html>

Sincerely,



Paul Van Caesele, MD FRCP
Medical Director, Cadham Provincial Laboratory
Chair, Manitoba Perinatal Screening Committee