Ontario Newborn Screening Laboratory
Children's Hospital of Eastern Ontario
Department of Genetics
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BULLETIN #4 – July 12, 2006

New disorders to be added to screening panel

As you know, since April 4, 2006, the Ontario Newborn Screening Laboratory has been screening for:

- Phenylketonuria (PKU) and Variants/Biopterin Defects (an Amino Acidemia)
- MCAD (medium chain acyl-CoA dehydrogenase) deficiency (a Fatty Acid Oxidation Defect [FAOD]), and
- Congenital hypothyroidism (an Endocrine disorder).

Beginning on August 8, 2006, you will start to see results for more disorders on the Screening Report, including results for other Amino Acidemias, other FAODs, as well as Organic Acidemias. All results will be reported as "positive" or "negative". Following are suggested test codes for your Laboratory Information Systems (LIS):

TEST CODES	TEST NAME (correspond to lines on the screening report)
NBS-PKU	Phenylketonuria and Variants / Biopterin Defects
NBS-MSUD	Maple Syrup Urine Disease
NBS-HCY	Homocystinuria (Hypermethioninemias)
NBS-CIT	Citrullinemias/Argininosuccinic Aciduria
NBS-TYR	Tyrosinemias
NBS-AA	Amino Acidopathies, other
NBS-C3	Propionic / Methylmalonic Acidemias
NBS-C5	Isovaleric Acidemia / 2 Methylbutyric Acidemia
NBS-C5DC	Glutaric Acidemia Type I
NBS-C5OH	3 Methylcrotonic / Hydroxymethylglutaric / Methylglutaconic / 2-Methyl,3-Hydroxybutyric acidemias, or B Ketothiolase Deficiency
NBS-OA	Organic Acidemias, other
NBS-MCAD	Medium Chain Acyl Dehydrogenase Deficiency/ Glutaric Acidemia Type 2
NBS-VLCAD	Very Long Chain Acyl Dehydrogenase Deficiency
NBS-LCHAD	Long Chain Hydroxy Acyl Dehydrogenase / Trifunctional Protein Deficiencies
NBS-CUD/CPT1	Carnitine Uptake Defect / CPT1 Deficiency
NBS-FA	Fatty acid oxidation disorders, other

Again, thank you for your cooperation and assistance in making the Ontario Newborn Screening Program the most effective and efficient that it can be.

Topics for future bulletins:

- new blood spot card and requisition
- transportation of samples

Previous bulletins (listed under 'Newsletter') and other information can be found at:
http://www.health.gov.on.ca/english/providers/program/child/screening/screen_sum.html

These bulletins are being circulated to keep you abreast of changes to the Ontario Newborn Screening program. Please share this information with any relevant personnel in your hospital / clinic. If there is someone who would like to be added to this list, contact: *Tillie Chiu, Genetic Counsellor*
(613) 738-3222, x1; NewbornScreening@cheo.on.ca