

## **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):

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