MARITIME Newborn Screening Dépistage néonatal DES MARITIMES

Conditions on the Maritime Newborn Screening Panel

The conditions screened for by Maritime Newborn Screening (MNBS) are rare and treatable. Working groups review potential conditions and the Advisory Committee makes recommendations about testing that would be most beneficial to the population it serves. The working groups and advisory committee include healthcare providers from the three Maritime Provinces and from a variety of specialty areas. Conditions can be nominated through the form on our website. Contact the clinical coordinator (phone: 902-470-2783) for more information.

Inherited Metabolic Disorders

Amino Acid Disorders

- Maple Syrup Urine Disease (MSUD)
- Phenylketonuria (PKU)
- Tyrosinemia Type I (TYR1)

Fatty Acid Oxidation Disorders

- Carnitine Palmitoyl Transferase 1 (CPT1) Deficiency
- Carnitine Palmitoyl Transferase 2 (CPT2) Deficiency
- Carnitine/Acylcarnitine translocase (CACT) Deficiency
- Carnitine Uptake Defect (CUD)
- Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
- Long Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency
- Trifunctional Protein (TFP) Deficiency
- Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency

Organic Acid Disorders

- Glutaric Acidemia Type I (GA1)
- Isovaleric Acidemia (IVA)
- Methylmalonic Acidemia (MMA): Cobalamin A & B Defects
- Methylmalonic Acidemia (MMA): Mutase Deficiency
- Propionic Acidemia (PA)

Urea Cycle Disorders

- Argininosuccinic Acidemia (ASA)
- Citrullinemia Type 1 (CIT)

Other Metabolic Disorders

- Classic Galactosemia
- Biotinidase Deficiency

Hemoglobinopathies

- Sickle Cell Disease (SS Disease (HbSS), SC Disease (HbSC), HbS-βthalassemia disease)
    - *Beta hemoglobin traits S, C, D, E, and unknown variants reported
- Alpha thalassemia disease (HbH Disease)
    - *Alpha thalassemia trait *not* reported

Endocrine Disorders

- Congenital Hypothyroidism (CH)

Other Disorders

- Cystic Fibrosis (CF)
- Severe Combined Immunodeficiency (SCID)
- Spinal muscular atrophy (SMA)