

## 1. Introduction

The goal of BC's Newborn Screening (NBS) Program is to identify babies who have a treatable disorder detectable through a blood test. These babies appear normal at birth and, unless they are screened, might otherwise not be diagnosed with one of these disorders before irreversible damage has occurred. If not treated, these conditions are associated with recurrent illnesses and/or developmental disabilities and/or death. Early detection of these disorders allows treatment that may prevent severe mental handicap, growth problems, health problems and sudden infant death.<sup>2</sup>

Babies born in British Columbia and the Yukon are screened for the following 24 disorders:

### Metabolic Disorders

Amino Acid Disorders:

- Phenylketonuria (PKU)
- Maple Syrup Urine Disease (MSUD)
- Citrullinemia (CIT)
- Argininosuccinic Acidemia (ASA)
- Homocystinuria (Hcy)
- Tyrosinemia 1 (Tyr 1)
- Guanidinoacetate Methyltransferase Deficiency (GAMT)

Fatty Acid Oxidation Disorders:

- Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
- Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
- Trifunctional Protein Deficiency (TFP)
- Very-Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
- Carnitine Uptake Disorder (CUD)

Organic Acid Disorders:

- Propionic Acidemia (PROP)
- Methylmalonic Acidemia (MUT)
- Cobalamin Disorders (Cbl A,B)
- Glutaric Aciduria Type 1 (GA1)
- Isovaleric Acidemia (IVA)

Galactosemia (GALT)

### Endocrine Disorders

- Congenital Hypothyroidism (CH)
- Congenital Adrenal Hyperplasia (CAH)

### Hemoglobinopathies

- Sickle Cell Disease (HbSS)
- Sickle Cell/Hemoglobin C (HbSC)
- Sickle Cell/ $\beta$ -thalassemia (HbS/ $\beta$ -thal)

### Cystic Fibrosis (CF)

Nine secondary disorders that are not primary targets of the screening program will be identified as “byproducts” of the screening process. See [Section 5, question 7](#), for a listing of these disorders.

Additional information on newborn screening for health care providers and families, including separate information sheets for each of the 24 disorders, is available on: [www.newbornscreeningbc.ca](http://www.newbornscreeningbc.ca) or [www.perinatalservicesbc.ca](http://www.perinatalservicesbc.ca).