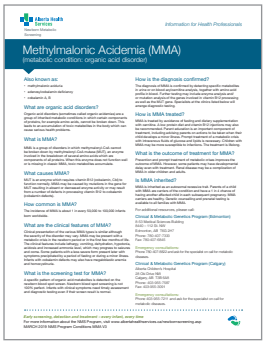
Methylmalonic Acidemia (MMA)
(metabolic condition: organic acid disorder)

Alberta Health Services
Newborn Metabolic Screening

Information for Health Professionals

Screened Conditions quick reference

Alberta's Newborn Metabolic Screening (NMS) Program provides accessible, current and standardized information for physicians and midwives.

Due to the time-sensitive nature of newborn blood spot screening, it is essential that physicians and midwives are able to access and generate information on the screened conditions in a straightforward manner.

What are the NMS Program condition fact sheets?

The condition fact sheets are a collection of resources for physicians and midwives that consist of information that describes the 21 treatable conditions screened for by the NMS Program.

They are

- evidence-informed
- user-friendly in terminology, language and format
- easily-accessible and understandable

Why are the NMS Program condition fact sheets important?

NMS Program condition fact sheets are important to help physicians and midwives understand rare conditions not often seen in regular practice. They also provide insight into next steps in the newborn blood spot screening pathway after an infant in their care has an abnormal screen result.

How are the NMS Program condition fact sheets used?

Physicians and midwives use the NMS Program condition fact sheets to guide next steps when an infant in their care has an abnormal screen result.

What do the NMS Program condition fact sheets cover?

The condition fact sheets cover the 21 treatable conditions that are screened for by the NMS Program.

Metabolic Conditions

- BIOT – Biotinidase deficiency
- CUD – Carnitine uptake defect
- CIT – Citrullinemia
- GA1 – Glutaric acidemia type 1
- GALT – Classic galactosemia
- HMG – 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency
- IVA – Isovaleric acidemia
- LCHAD – Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
- MCAD – Medium chain acyl-CoA dehydrogenase deficiency
- MMA – Methylmalonic acidemia
- MSUD – Maple syrup urine disease
- PA – Propionic acidemia

- PKU – Phenylketonuria
- TFP – Tri-functional protein deficiency
- TYR1 – Tyrosinemia type 1
- VLCAD – Very long chain acyl-CoA dehydrogenase deficiency

Endocrine Conditions

- CAH – Congenital adrenal hyperplasia
- CH – Congenital hypothyroidism

Cystic Fibrosis (CF)

Sickle Cell Disease (SCD)

- SCT – Sickle cell trait

Severe Combined Immunodeficiency (SCID)

Each NMS Program condition fact sheet gives information about

- the condition
- the incidence rate
- clinical features
- the screening test
- confirming the diagnosis
- treatment and management
- where to find more information

How are the NMS Program condition fact sheets accessed?

The NMS Program condition fact sheets can be accessed through and printed from www.ahs.ca/newbornscreening

The NMS Lab faxes the appropriate condition fact sheet to physicians and midwives who have an infant in their care that has an abnormal screen result.