



Inborn Errors of Metabolism Detectable by Extended Newborn Screening by LC-MSMS

The following is a list of disorders in the screening panel that are potentially detectable by tandem mass spectrometry (MS/MS). However due to the nature of the disorders and the existence of milder and variant forms, not all individuals with these disorders will be identified by newborn screening. Please note that although this list includes those disorders most likely to be detected it is not exhaustive and in the course of investigating abnormal results other conditions may be detected.

Normal screening results indicate that the individual has a very low risk of having one of the listed disorders but does not completely rule out the possibility. Where there is clinical concern a metabolic physician should be consulted even in the face of normal screening result. Abnormal screening results indicate a higher risk for one of the above listed disorders and the child will need to be seen by a metabolic physician for evaluation and diagnostic testing.

Please also note that there are many inborn errors of metabolism that cannot be detected by this extended newborn screen. Where there is clinical concern, a metabolic physician should be consulted.

Primary Targets

Those conditions the programme is aiming to detect

Amino Acid Disorders:

- Phenylketonuria including bipterin defects
- Maple Syrup Urine Disease
- Citrullinaemia type 1
- Argininosuccinic Aciduria
- Tyrosinaemia type 1
- Homocystinuria (pyridoxine unresponsive)

Organic Acid Disorders:

- Propionic acidaemia
- Methylmalonic acidaemia (MUT)
- Cobalamin A/B
- Isovaleric acidaemia
- β -ketothiolase deficiency
- Glutaric acidaemia type 1
- Malonic aciduria
- 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency
- Multiple carboxylase deficiency

Fatty Acid Oxidation Disorders:

- Primary carnitine deficiency / Carnitine uptake deficiency
- Medium chain acyl-CoA dehydrogenase deficiency
- Very long chain acyl-CoA dehydrogenase deficiency



- Long chain hydroxy acyl-CoA dehydrogenase
- Trifunctional protein deficiency

Secondary Conditions

Conditions that may well be identified while investigating abnormal results for the primary targets.

Amino Acid Disorders:

- Hyperphenylalanine
- Argininase deficiency
- Citrin deficiency
- Hypermethioninaemia
- Tyrosinaemia Types 2 and 3

Organic Acid Disorders:

- 3-Methylcrotonyl-CoA carboxylase deficiency
- 2-Methyl-3-hydroxy butyric aciduria
- 3-Methylglutaconyl-CoA dehydratase deficiency
- Isobutyryl-CoA dehydrogenase deficiency
- 2-Methylbutyryl-CoA dehydrogenase deficiency/ short branch chain acyl-CoA
- Ethylmalonic encephalopathy
- Cobalamin C/D

Fatty Acid Oxidation Disorders:

- Carnitine palmitoyltransferase deficiency type 1
- Carnitine palmitoyltransferase deficiency type 2
- Carnitine-acylcarnitine translocase deficiency
- Multiple acyl-CoA dehydrogenase deficiency / glutaric aciduria type 2
- Short chain acyl-CoA dehydrogenase deficiency
- Medium/Short chain hydroxy acyl-CoA dehydrogenase deficiency
- Medium chain ketoacyl-CoA thiolase deficiency