

# Newborn Metabolic Screening Programme (NMSP): Expanded Screening

November 2006

---

From 1 December 2006, the NMSP will expand the number of metabolic disorders that are screened for in the programme. This is possible due to the Starship Foundation gifting a new machine called a tandem mass spectrometer or 'tandem'.

Australia and the United States have been using the technology since 1998. The tandem allows New Zealand babies to be tested for the same disorders as Australian babies.

The new disorders are grouped as follows:

## **1. Disorders of amino acid breakdown (about 15 conditions) – examples include:**

- Citrullinaemia
- Glutaric acidaemia
- Methylmalonic acidemia

Each disorder is caused by a missing enzyme. Without the enzymes, waste products such as ammonia rise to harmful levels. We already screen for two amino acid breakdown disorders – PKU and MSUD.

The treatment for these disorders is a special diet – for instance for PKU, special diet restrictions to reduce protein (and therefore phenylalanine in the diet) and supplemental additions of vital amino acids.

## **2. Disorders of fatty acid oxidation (about 10 conditions) – examples include:**

- Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
- Carnitine transporter defect

Each disorder is caused by a missing enzyme. Without these enzymes, energy cannot be used from fats. Without energy from fats, the body can run out of energy.

The treatment for these disorders is regular feeding and close supervision when the individual is ill. For instance, if a baby has vomiting and/or diarrhoea, that they are constantly monitored and have adequate food intake.

- There is no change to the amount of blood required.
- The blood card remains the same.
- The sample should be taken at 48 hours or as soon as possible thereafter.
- Please post the sample to the laboratory on the day of sample taking.

## Full list of new conditions

### Disorders of amino acid breakdown:

- Argininaemia/arginase deficiency
- Argininosuccinic aciduria (ASA lyase deficiency)
- Citrullinaemia (argininosuccinate synthase deficiency, citrin deficiency)
- Fumaryl acetoacetase deficiency (Tyrosinaemia Type I)
- Homocystinuria (cystathionine beta-synthase deficiency)
- Pterin defects
- Tyrosine aminotransferase deficiency (Tyrosinaemia Type II)
- Beta-ketothiolase deficiency (mitochondrial acetoacetyl-CoA thiolase deficiency)
- Cobalamin C defect (homocystinuria with methylmalonic aciduria)
- Glutaryl CoA dehydrogenase deficiency (glutaric acidaemia type I)
- Holocarboxylase synthetase deficiency
- 3-hydroxy-3-methylglutaryl-CoA lyase (HMGCoA lyase) deficiency
- Isobutyryl-CoA dehydrogenase deficiency
- Isovaleric acidaemia
- Methylmalonic acidurias (mutase deficiency, CblA and CblB defects)
- Propionic acidaemia
- 3-methylcrotonyl-CoA carboxylase deficiency
- 2-methylbutyryl-CoA dehydrogenase deficiency
- 3-methylglutaconyl-CoA hydratase deficiency
- Maternal vitamin B12 deficiency

### Disorders of fatty acid oxidation:

- Carnitine/acylcarnitine translocase deficiency
- Carnitine transporter defect
- CPT-1 (carnitine palmitoyl transferase deficiency type I)
- CPT-2 (carnitine palmitoyl transferase deficiency type II)
- LCHAD (3-hydroxy long chain acyl-CoA-dehydrogenase deficiency)
- MCADD (medium chain acyl-CoA-dehydrogenase deficiency)
- MADD (multiple acyl-CoA-dehydrogenase deficiency)
- SCHAD (short chain hydroxy acyl-CoA-dehydrogenase deficiency)
- TFP (trifunctional protein deficiency)
- VLCAD (very long chain acyl-CoA-dehydrogenase deficiency)