

Information for parents and carers about the new test for rare metabolic disorders

November 2006

What is newborn metabolic screening?

When talking with your Lead Maternity Carer (LMC), you will be offered a screening test for your baby which detects rare but life threatening metabolic disorders. If you consent to this test, a blood sample will be taken from the heel of your baby at about 48 hours of age. The blood is put on a card (often called a "Guthrie" card) and sent to the National Testing Centre at Auckland City Hospital to be screened for these disorders.

For many years, babies have been screened for seven disorders: phenylketonuria (PKU), congenital hypothyroidism, cystic fibrosis (CF), congenital adrenal hyperplasia (CAH), galactosaemia, biotinidase deficiency and maple syrup urine disease (MSUD). All of these conditions are best detected and treated early to avoid illness and death.

What are the new disorders?

With new technology, from December 1 2006, New Zealand babies will have the same screening as Australian babies. This new technology, called a tandem mass spectrometer, allows for testing of several metabolic disorders, using the same amount of blood.

These rare disorders can be divided into two groups - disorders of amino acid and fatty acid metabolism. The amino acid disorders are similar to phenylketonuria (PKU) and maple syrup urine disease (MSUD). The most common fatty acid disorder is Medium Chain Acyl-CoA Dehydrogenase Deficiency or MCADD.

What is the cause of these disorders?

These disorders are genetic disorders caused by an enzyme deficiency. Enzymes are chemicals which are essential to the body's metabolism. The new tests screen for some enzyme deficiencies that affect how the body can use proteins and fats. If there is an enzyme deficiency, there can be a build up of an amino acid (the building blocks of protein) or a carnitine (attached to fat) in the blood and these are the substances we test for.

What if the results are positive?

Newborn metabolic screening currently finds about 35 babies each year that have one of the disorders. These babies are referred to paediatricians or a metabolic specialist for further diagnostic testing for confirmation of a disorder and for treatment. About 5 - 10 more babies a year will be found to have one of these disorders. These are babies for whom early treatment and advice is vital to prevent illness.

Not all babies with a positive screening result have something wrong. If a result is clearly abnormal, newborn screening staff will telephone the baby's LMC. If there is a borderline result, the newborn screening team may ask for a second blood sample from your baby. Most repeat samples have normal results.

What is the treatment for these disorders?

The disorders are treatable with either dietary management or medication. If your baby has one of these disorders, you will be asked to come and see a paediatrician or metabolic specialist who will be able to answer all your questions and advise about treatment.

If you would like more specific information, please phone the National Testing Centre on:
tel (09) 307 4949 ext 23019.