



INFORMATION FOR PARENTS AND CARERS ABOUT THE NEW TEST FOR OVER 20 RARE METABOLIC DISORDERS

What is Newborn Screening?

Your baby had a blood sample taken from the heel at about 3 days of age. The blood was absorbed into a special card and sent to the NSW Newborn Screening Laboratory in Sydney where it was tested for several disorders.

For many years all babies have been tested for four disorders: Congenital Hypothyroidism, Cystic Fibrosis, Phenylketonuria and Galactosaemia. Early treatment is important.

A new test for very rare disorders

Now Newborn Screening is using new technology (tandem mass spectrometry) to test for 20 or more rarer metabolic disorders, using the same very small quantity of blood.

What are the rarer metabolic disorders?

Rare disorders of amino acid metabolism, of organic acid metabolism, and of fatty acid metabolism. These disorders include such things as maple syrup urine disease, methylmalonic aciduria, medium chain acyl CoA dehydrogenase deficiency, and many more.

What causes the rarer metabolic disorders?

These disorders are genetic disorders caused by an enzyme deficiency. Enzymes are chemicals which are essential for 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 result is positive?

Newborn Screening finds about 90 babies each year that have one or other of the disorders. These babies are referred to paediatricians or clinical geneticists for further diagnostic testing for confirmation of a disorder and to start treatment.

About 10-12 of these babies have one of the rarer metabolic disorders. These are babies for whom early treatment and advice is vital to prevent illness.

Not all babies with a positive test result have anything wrong. If a result is clearly abnormal, newborn screening staff will telephone the baby's doctor. If there is a borderline result Newborn Screening may ask for a second blood sample from your baby for one of the rarer metabolic disorders. **MOST REPEAT SAMPLES HAVE QUITE NORMAL RESULTS.**

Treatment

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 genetic metabolic specialist who will be able to answer all your questions and advise about treatment. If your family lives in a country area of New South Wales, it may be possible for you to see a paediatrician closer to your town.

If you need any more specific information please telephone the Newborn Screening Laboratory on (02) 9845 3255 or (02) 9845 3659.

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For further information please contact your paediatrician or the NSW Newborn Screening Programme
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