



INFORMATION FOR PARENTS AND CARERS

RESAMPLE NEWBORN SCREENING TEST FOR GALACTOSAEMIA

What is the Newborn Screening Programme?

A blood sample was taken from the heel at about three days of age. The blood was absorbed into a special card, dried, and sent to the Newborn Screening Laboratory in Sydney where it was tested for several disorders. Galactosaemia was one of the disorders.

Why are babies screened for Galactosaemia?

Galactosaemia is an extremely rare disorder. The incidence of true galactosaemia is 1:50,000 or about 2 babies a year in New South Wales. Babies are screened for galactosaemia so that they can be treated early.

Why does my baby need a second test for Galactosaemia?

The test result from your baby's first sample was slightly abnormal and the laboratory

needs a repeat sample, usually when your baby is about one month old, to make sure your baby does not need treatment. There are mild variant forms of galactosaemia for which there are not any adverse effects and for which most babies have **normal results when retested**. These babies do not have galactosaemia and are **not** lactose intolerant.

What is galactosaemia?

Galactosaemia is an enzyme deficiency. It occurs when both parents pass a galactosaemia gene to their baby.

Galactosaemia causes an accumulation in the blood of galactose, a sugar which is in milk. The enzyme deficiency prevents the galactose from being changed to another sugar, glucose. Galactose and other compounds accumulate and cause lethargy, vomiting, jaundice and sepsis.

Babies with galactosaemia need treatment to avoid these problems. The treatment is a diet which completely excludes galactose. For newborn babies this means a special formula.

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