NEWBORN SCREENING TEST FOR GALACTOSAEMIA
INFORMATION FOR PARENTS AND CARERS

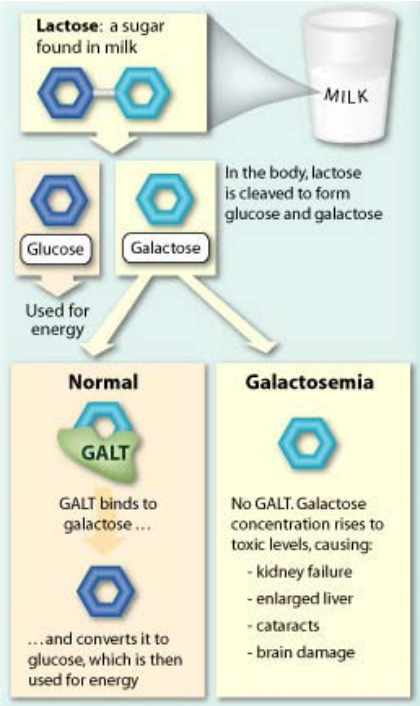
What is the Newborn Screening Programme?

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 Potchefstroom (Ampath and North-West University Newborn Screening Programme) 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. Babies are screened for galactosaemia so that they can be treated early.

What is galactosaemia?



Galactosemia is a rare disorder that affects the body's ability to break down a food sugar called galactose (found in milk and other dairy products).

Normally, the body breaks down lactose into galactose and then into glucose (a sugar used for energy). People with galactosemia are missing an enzyme called GATL (galactose-1-phosphate uridyl transferase), which normally converts galactose into glucose. Without this enzyme, harmful amounts of galactose build up in the blood.

Interesting facts about galactosemia

Galactosemia was first discovered in 1908 by the physician Von Ruess. Classical galactosemia affects 1 in every 55,000 newborns.

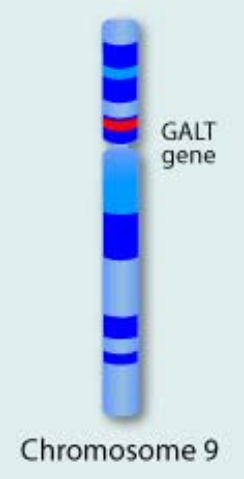
What are the symptoms of galactosemia?

The build-up of galactose in the body can cause several severe symptoms: kidney failure, an enlarged liver, cataracts (clouding of the eye lens), poor growth, and mental retardation.

People can inherit a milder form of the disorder when a different gene, also involved in galactose metabolism, is mutated. These patients often suffer from cataracts, but not the other symptoms associated with classical galactosemia.

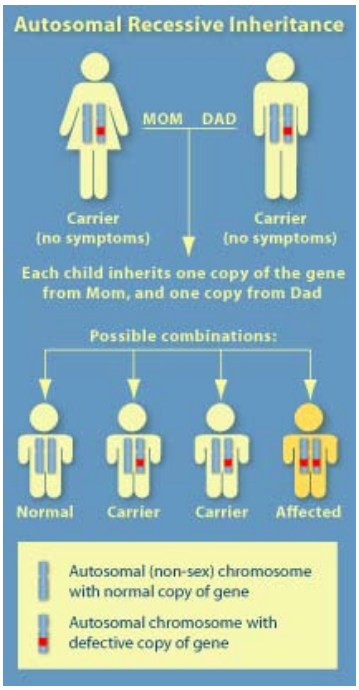
How do people get galactosemia?

Tel: 018-299 2310/293 9510 | Fax: 018-299 2316 | E-mail: NBS@Ampath.co.za
Address: Biochemistry, NWU, Private Bag X6001, Potchefstroom, 2520
www.newbornscreening.co.za OR www.ampath.co.za/newborn_screeningIndex.htm



The most common form of the disorder, classic galactosemia, is passed down in an autosomal recessive pattern. To get the disorder, a child must inherit one defective gene from each parent. Inheriting one normal gene and one mutated gene makes a person a carrier. A carrier produces less of the GALT enzyme than normal, but is still able to break down glucose and avoid having symptoms of galactosemia. However, carriers can still pass on the mutated gene to their children.

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



How is galactosemia treated?

Babies with galactosaemia need treatment to avoid these problems. The only way to treat galactosemia is through dietary restrictions. People with the disorder must stay away from foods and drinks containing galactose, including milk, cheese, and legumes (dried beans). For newborn babies this means a special formula.

How do doctors diagnose galactosemia?

In most states, babies are tested for galactosemia at birth. Using a tiny blood sample taken from the baby's heel, the test checks for low levels of the GALT enzyme. This allows for prompt treatment, which can substantially prevent the serious symptoms of this disorder.

For those families with a history of the disorder, a doctor can determine during a woman's pregnancy whether her baby has galactosemia 1) by taking a sample of fluid from around the fetus (amniocentesis), or 2) by taking a sample of fetal cells from the placenta (chorionic villus sampling or CVS

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 galactosemia for which there are not any adverse effects and for which most babies have normal results when retested. These babies do not have galactosemia and are not lactose intolerant.

Tel: 018-299 2310/293 9510 | Fax: 018-299 2316 | E-mail: NBS@Ampath.co.za
Address: Biochemistry, NWU, Private Bag X6001, Potchefstroom, 2520
www.newbornscreening.co.za OR www.ampath.co.za/newborn_screeningIndex.htm