



State of CT Genetics Newborn Screening Program Family Fact Sheet

GALACTOSEMIA

Carbohydrates are important parts of our diet. Babies who have the disorder called galactosemia (Guk-LAK-toe-SEE-me-ah) cannot break down certain parts of carbohydrates called galactose. Galactose, is a sugar, found in milk, formula, breast milk, and other foods. Babies with galactosemia have high levels of galactose in their body. If galactosemia is not treated, the high levels of galactose can cause mental retardation, eye disease, liver disease, and other physical problems.

Connecticut began screening for galactosemia in 1964. In Connecticut, about 1 in every 47,540 newborn babies has galactosemia.

How does a baby get galactosemia?

Galactosemia is an inherited condition. Both the mother and the father have one altered gene that does not produce the enzyme necessary to break down the carbohydrate, galactose. A baby has to receive this gene from both the mother and the father to have this condition.

How is a baby tested for galactosemia?

Prior to discharge from the facility of birth, a small sample of blood is taken from the baby's heel. The sample is then sent to the State laboratory to screen for galactosemia. Abnormal screening results are reported to the Department of Public Health, Genetic Newborn Screening Tracking Unit nurse consultant. The DPH nurse consultant will notify the baby's primary care provider of the need for a prompt referral to one of the Genetic Regional Treatment Centers at Yale or UCONN for confirmation testing and follow-up treatment if necessary.

What is the treatment for galactosemia?

The Regional Treatment Center specialists will prescribe treatment for galactosemia if necessary, which may include a special milk free diet. Early diagnosis of galactosemia leads to early treatment, which is important for the health of your baby.



Keeping Connecticut Healthy

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