Duarte Galactosemia (DG)

What is newborn screening?

Newborn blood spot screening is a screening test to check for health conditions that do not show symptoms at birth. Soon after birth, a nurse takes a few drops of blood from your baby's heel. Newborn screening helps to detect many conditions before symptoms start to show.

KEY POINTS:

- **Duarte galactosemia is different from classic galactosemia.**
- **DG is considered by most healthcare professionals to be clinically mild.**
- **Most individuals with DG never develop any health problems.**
- **Many specialists do not recommend any treatment or diet change for DG.**

What was found on the newborn screen?

The newborn screen that was collected at birth found enzyme levels that were suggestive of a condition called galactosemia. Galactosemia is a genetic disorder (present at birth) where the body is not able to use a milk sugar called galactose. Galactose comes from food, including all breast milk, all dairy products, and many baby formulas. Your baby had follow up testing to see if he/she has galactosemia.

What were the results of follow up testing?

Follow up testing showed that your baby has a condition called **Duarte galactosemia (DG).** DG is a more common and far less severe form of galactosemia. DG occurs when babies have smaller amounts of and/or weaker forms of the GALT enzyme. The GALT enzyme help the body break down and use galactose sugars. **Most individuals with DG never develop any symptoms and require no treatment.**

How is Duarte Galactosemia different from Classic Galactosemia?

Duarte galctosemia is different from the classic form of galactosemia because patients with DG have enough GALT enzyme to help with breaking down galactose. This is different from patients with classic galactosemia because they have zero or very little GALT enzyme.

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What causes Duarte Galactosemia?

DG is a genetic condition; this means it is inherited through the genes we get from our parents. DG occurs when an enzyme, called galactose-1 phosphate uridyl transferase (GALT) is not working at full strength. This enzyme's job is to help change galactose into glucose.

What health problems can it cause?

Most individuals with DG never develop health problems due to DG. While specialists were still learning about DG, they used to give the same recommendations they used in people with classic galactosemia: to avoid dairy that had galactose. However, research has shown that this is not needed. There has been no differences in developmental outcomes between children with DG and children without DG, even if they were exposed to cow's milk or dairy.

So treatment isn't needed?

Based on recent studies, many specialists no longer recommend any diet change for babies with DG. Unless told otherwise, you can continue to breastfeed and/or formula feed as you have been.

What if I still have questions?

We understand that this can be an overwhelming and emotional process. Many families have questions and concerns. The Connecticut Newborn Diagnosis and Treatment Network (the Network) is available to put you in touch with the best resource and/or to connect you with a genetic counselor. To reach the Network, you can call 860-837-7870, Monday-Friday, 8:30am-4:30pm. We also recommend the following websites as accurate and informative resources:

www.babysfirsttest.org

www.duartegalactosemia.org

www.newbornscreening.info

This fact sheet was written for information purposes only.
It should not replace medical advice, diagnosis or treatment.

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