

Commonly asked questions

1. What is galactosemia?

Galactosemia is a disorder caused by excess of galactose, a milk sugar, in the body. The defect is in one of the enzymes responsible for converting galactose to glucose that can be used by the body for fuel. Excess galactose products damage the liver, eyes, kidneys, and brain and can cause life-threatening infection in infants.

2. How and when will we know if my baby has galactosemia?

If your baby's newborn screening result showed a GAL level > 13 mg / dl, he or she probably has galactosemia. If the result was 6 - 13 mg /dl your baby either could still have galactosemia or a harmless variant. The newborn screening test will be repeated and additional tests will be undertaken to help determine if your baby has galactosemia or not. Typically the results of these tests take up to 4 days to come back. Depending on the test results, additional testing can take a variable amount of time to confirm the diagnosis. In a very small minority of cases, it can be difficult to determine whether a child is affected or not.

3. How did my baby get this?

Galactosemia is an autosomal recessive disorder. This means that your baby has two mutated genes, one from the mother and one from the father. Having only one mutated gene (a carrier) does not affect a person at all. Similarly having one mutated gene together with a common variant, referred to as 'Duarte galactosemia', does NOT appear to affect the individual at all.

4. What does it mean for my child?

If your baby has galactosemia, he or she will have to stay on a milk free diet throughout life. This will prevent the life threatening infections, mental retardation, liver and kidney disease and development of cataracts. Most children with galactosemia do however have specific difficulties, particularly with speech and girls develop premature ovarian failure despite early treatment. However, specific early intervention & speech therapy is very helpful.

5. What is the treatment? Does it work? Is the diet difficult to do/expensive?

Galactosemia is primarily treated by a milk free diet. Therefore infants with galactosemia cannot be allowed to have lactose / galactose containing foods such as breast milk or regular milk formula but this is safely replaced by soy formulas. This diet is very effective at preventing the life threatening complications of galactosemia. It is less convenient than regular diet but most people find soon it becomes relatively routine and is not significantly more expensive.

6. What about my other children/future children?

As galactosemia is an inherited recessive genetic condition it is important to have your other children tested. Children from the same father and mother as the affected infant have a 1 in 4 (25%) chance of having galactosemia though it is unlikely if they had no problems in infancy.

Since there is a risk for having a future child with galactosemia it is important to let your obstetrician and pediatrician know that you have a child with galactosemia if you are planning future pregnancies so that they may discuss the options with you and prepare accordingly.