A Parent Guide

August 2009

Galactosemia Variations

This information sheet will summarize the genetics of galactosemia and explain some of the issues involved when your child is identified as having a galactosemia allele variant.

WHAT IS GALACTOSEMIA?

Galactosemia is an inherited condition. Every person is born with a number of enzymes, or proteins, that are needed to break down fats and sugars for energy. One of those enzymes, galactose-1-phosphate uridyltansferase or GALT, is important in the break down of a specific sugar called galactose. Galactose is one of the main components of lactose, the primary sugar found in milk and dairy products. People with galactosemia have a deficiency of the GALT enzyme. This means that their GALT enzyme does not adequately break down galactose, which accumulates in the body as galactose-1-phosphate can lead to health problems.

Classical galactosemia is diagnosed in approximately one out of every 50,000 babies. These babies have very little to no working GALT enzyme. They can have jaundice, liver problems, vomiting, poor feeding, failure to grow, infections, speech disabilities, mental handicaps and cataracts of the eye. Treatment includes a specialized diet with limited galactose products.

There are other variations of the galactosemia gene. Most commonly seen are Duarte (a low enzyme producing gene) and Los Angeles (an enhanced enzyme producing gene). These variants and combination of genes (gN, DN, DD, LAN, LAg, LAD) do not require treatment or dietary restriction of galactose. This table describes the genes seen in Utah. The activity level shows the efficiency of the GALT enzyme.

Gene	Symbol	Activity level
Normal	N	100%
Galactosemia - non-functioning	g	0%
Duarte - partially functioning	D	50%
Los Angeles - above normal functioning gene	LA	125%

The **Duarte variant** is found in 1 in 20 persons. Babies who have one or more Duarte alleles have some GALT enzyme (10-25% activity) and do not commonly have any symptoms. Recent studies have shown that children with Duarte galactosemia who continue to receive breast milk in the first year of life might have higher than normal galactose-1-phosphate levels, but they do not have any symptoms or health problems related to the Duarte variant.

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Newborn Screening Program

The **Los Angeles** variant is found in approximately 1 in 30 persons. The Los Angeles allele has more than the normal GALT enzyme (125% activity) and does not cause symptoms or health problems.

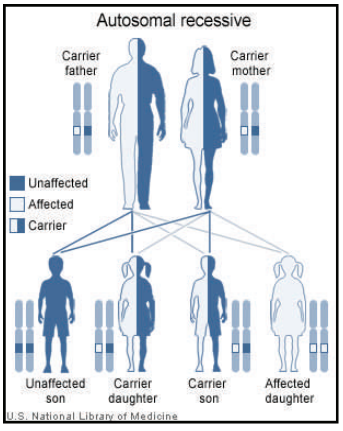
Each person has a different combination of GALT genes or variants in their unique gene pair, depending on which gene they inherited from each parent. Different combinations in a gene pair result in different levels of GALT enzyme activity in the body. This table shows the possible variations in the gene pair and their activity . Only the classical form needs treatment.

	Genotype (gene pair)	Diagnosis	GALT Enzyme Activity	GALT Enzyme Function	Clinical Management
2 normal genes	NN	Normal	100%	Normal	None
2 nonfunctioning genes	gg	Classical Galactosemia	0%	Absent	Diet, specialized medical care
2 Duarte genes	DD	Homozygous Duarte Variant	50%	Reduced	None
Combinations:	Ng	Galactosemia Gene Carrier	50%	Reduced	None
	ND	Duarte Variant Carrier	75%	Slightly reduced	None
	Dg	Duarte Galactosemia	5-20%	Reduced	None
	NLA	LA Variant Carrier	112%	Above normal	None
	LAg	Galctosemia Gene Carrier	62%	Slightly reduced	None

IS GALACTOSEMIA A GENETIC CONDITION?

Yes. Almost all of our genes come in pairs, one inherited from each of our parents. (Refer to diagram.)

A carrier of a genetic condition is a person who has one copy of an altered gene. A recessive condition, like classical galactosemia, results when a child inherits two copies of an altered gene, one from each parent. Both parents must be carriers of the same recessive disease gene in order for any of their children to have the condition.



Carriers themselves do not have the condition. The only consequence of being a carrier of a recessive gene is the possibility of transmitting that particular gene to a child. Carriers have a 1-in-2 chance with each pregnancy to transmit the altered gene to future children.

We do know that each time a baby is diagnosed with a g, D or LA gene, his or her parents have a 1-in-4 (or 25%) chance for each of the next children to also be diagnosed with that same condition (the diagnosis of galactosemia for each child is independent from his or her siblings).

A genetic counselor is available for education for you and for your child before his/her child bearing years. Genetic counseling and education may be obtained through the Division of Medical Genetics, at the University of Utah, (801) 585-2457.