Understanding Type 1 Galactosemia

Type 1 galactosemia is a rare, progressive genetic disease that can be life-threatening for newborns and can cause lifelong cognitive, neurological, speech, and fertility complications.1 Type 1 galactosemia is caused by mutations in the GALT gene.2 The GALT gene’s job is to tell the body’s cells how to make the galactose-1-phosphate uridylyltransferase enzyme, or the GALT enzyme.1,2

- Type 1 galactosemia includes classic, clinical variant, and Duarte galactosemia1
- Symptoms of Type 1 galactosemia typically occur in classic and clinical variant galactosemia1

Why do we need GALT enzymes?

GALT enzymes help the body process galactose, a sugar that is naturally produced in the body and found in dairy, breast milk, and other foods.2,3 Genetic mutations in the GALT gene mean the body is not able to produce enough of the GALT enzyme.1,2

Without the GALT enzyme, a buildup of galactose occurs, which results in the accumulation of two additional substances called galactose-1-phosphate (Gal-1p) and galactitol. Too much galactose, Gal-1p, and galactitol in the body is toxic and can contribute to lifelong complications.2,3

The risk and severity of galactosemia depends on the types of mutations in the GALT gene and the level of GALT enzyme activity.1

Restricting galactose in people with galactosemia is critical, but not enough.

The current standard of care for galactosemia is a galactose-restricted diet.3 If galactose is removed from a newborn’s diet quickly, severe symptoms, like liver failure and death may be avoided.2

However, the body produces endogenous galactose, which means it produces galactose naturally, no matter what foods are eaten.1 So, even when the recommended diet is strictly followed, lifelong complications can still occur.2

Lifelong Complications

As children with Type 1 galactosemia grow, the buildup of galactose, Gal-1p and galactitol continues, which can contribute to lifelong complications.2,3

- Cognitive and motor delays (49.7%)4
- Neurological complications (52%), with tremors as the most frequent complication (31%)4
- Language delay (78%), and language and speech disorders (66.4%)4
- Primary ovarian insufficiency (79.7%), which often requires hormone replacement therapy (83.5%)4 and may lead to difficulty in getting pregnant
- Poor growth5 and delayed puberty for girls (48.5%) and boys (4.8%)4
- Movement and coordination challenges (27%)4
- Cataracts (25.8%)4

The numbers above represent the percentage of people with Type I galactosemia who reported symptoms in a study of 509 participants.4
How is a child born with galactosemia?

Galactosemia is an autosomal recessive disease. This means the parents of a child with galactosemia are usually carriers of the genetic mutation. Carriers have one dominant working gene and one recessive non-working gene with a mutation. Carriers do not experience symptoms of the disease. **A child is born with galactosemia when they inherit two copies of the gene mutation.**

Newborn Screening

Because galactosemia is life-threatening, newborn screening for Type 1 galactosemia is conducted in every state in the United States and several other countries.

Newborn screening is usually done within 48 hours of birth; however, the results may not be available for several days. Since many parents are unaware that they are carriers, a child may be fed breastmilk or infant formula containing galactose, which can cause life-threatening complications.

References