GALE Gene

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UDP-galactose-4-epimerase

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1. Normal Function

The *GALE* gene provides instructions for making an enzyme called UDP-galactose-4-epimerase. This enzyme enables the body to process a simple sugar called galactose, which is present in small amounts in many foods. Galactose is primarily part of a larger sugar called lactose, which is found in all dairy products and many baby formulas.

UDP-galactose-4-epimerase converts a modified form of galactose (UDP-galactose) to another modified sugar (UDP-glucose). Glucose is a simple sugar that is the main energy source for most cells. This enzyme also promotes the reverse chemical reaction, the conversion of UDP-glucose to UDP-galactose. UDP-galactose is used to build galactose-containing proteins and fats, which play critical roles in chemical signaling, building cellular structures, transporting molecules, and producing energy.

2. Health Conditions Related to Genetic Changes

2.1 Galactosemia

More than 20 mutations in the *GALE* gene have been identified in people with a form of galactosemia known as type III or galactose epimerase deficiency. The signs and symptoms of this condition begin shortly after birth and can vary from mild to severe. Most of the genetic changes alter a single protein building block (amino acid) in UDP-galactose-4-epimerase, which makes the enzyme unstable or impairs its usual function.

Some *GALE* gene mutations severely reduce or eliminate the activity of UDP-galactose-4-epimerase in all of the body's tissues. These genetic changes lead to a severe form of galactosemia type III described as the generalized form. A loss of enzyme activity prevents cells from processing galactose obtained from the diet. As a result, compounds associated with galactose processing can build up to toxic levels in the body. The accumulation of these substances damages tissues and organs, leading to serious complications such as clouding of the lens of the eye (cataract), intellectual disability, and damage to the liver, kidneys, and brain.

Other mutations in the *GALE* gene reduce the activity of UDP-galactose-4-epimerase in red blood cells only. These genetic changes underlie a much milder form of galactosemia type III described as the peripheral form. Affected individuals may not have any of the complications typically associated with galactosemia and often do not require treatment. Researchers are unclear why the effects of some *GALE* mutations are restricted to blood cells, while other mutations affect all of the body's tissues and cause severe health problems.

3. Other Names for This Gene

- galactowaldenase
- GALE_HUMAN
- SDR1E1
- UDP - Uridyl diphosphate galactose-4-epimerase
- UDP Galactose Epimerase

- UDP-Glucose 4-Epimerase
- Uridine diphosphate galactose-4-epimerase
- Uridine Diphosphate Glucose Epimerase

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