GALK1 Gene

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Galactokinase 1

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1. Normal Function

The *GALK1* gene provides instructions for making an enzyme called galactokinase 1. 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.

Galactokinase 1 is responsible for one step in a chemical process that converts galactose into other molecules that can be used by the body. Specifically, this enzyme modifies galactose to create a similar molecule called galactose-1-phosphate. A series of additional steps converts galactose-1-phosphate to another simple sugar called glucose, which is the main energy source for most cells. Galactose-1-phosphate can also be converted to a form that is used to build galactose-containing proteins and fats. These modified proteins and fats 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 30 mutations in the *GALK1* gene have been identified in people with a form of galactosemia called type II or galactokinase deficiency. Affected infants develop clouding of the lens of the eye (cataracts) but otherwise experience few of the long-term complications associated with classic galactosemia. Most of these mutations change single protein building blocks (amino acids) in galactokinase 1. A few mutations delete a small amount of genetic material from the *GALK1* gene, resulting in an unstable or inactive version of this enzyme.

A shortage of functional galactokinase 1 prevents cells from processing galactose obtained from the diet. As a result, galactose and a related sugar called galactitol can build up, particularly in cells that make up the lens of the eye. An accumulation of these substances damages the lens, causing it to become cloudy and leading to blurred vision.

3. Other Names for This Gene

- ATP:D-galactose 1-phosphotransferase
- galactokinase
- GALK
- GALK1_HUMAN
- GK1

References

1. Berry GT. Classic Galactosemia and Clinical Variant Galactosemia. 2000 Feb 4[updated 2020 Jul 2]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH,Stephens K, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): Universityof Washington, Seattle; 1993-2020. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK1518/

2. Holden HM, Thoden JB, Timson DJ, Reece RJ. Galactokinase: structure, function and role in type II galactosemia. Cell Mol Life Sci. 2004 Oct;61(19-20):2471-84. Review.
3. Thoden JB, Holden HM. The molecular architecture of humanN-acetylgalactosamine kinase. J Biol Chem. 2005 Sep 23;280(38):32784-91.
4. Timson DJ, Reece RJ. Functional analysis of disease-causing mutations in humangalactokinase. Eur J Biochem. 2003 Apr;270(8):1767-74.
5. Timson DJ, Reece RJ. Sugar recognition by human galactokinase. BMC Biochem.2003 Nov 4;4:16.
6. Timson DJ. The molecular basis of galactosemia - Past, present and future.Gene. 2016 Sep 10;589(2):133-41. doi: 10.1016/j.gene.2015.06.077.
