GCK gene

glucokinase

Normal Function

The *GCK* gene provides instructions for making a protein called glucokinase. This protein plays an important role in the breakdown of sugars (particularly glucose) in the body. Glucokinase is primarily found in the liver and in beta cells in the pancreas. Beta cells produce and release (secrete) the hormone insulin, which helps regulate blood glucose levels by controlling how much glucose is passed from the bloodstream into cells to be used as energy. Glucokinase acts as a sensor, recognizing when the level of glucose in the blood rises and helping stimulate the release of insulin from beta cells to control it. In the liver, glucokinase helps determine when excess glucose should be taken in and converted to glycogen, which is a major source of stored energy in the body.

Health Conditions Related to Genetic Changes

Maturity-onset diabetes of the young

Mutations in the *GCK* gene cause maturity-onset diabetes of the young (MODY), which is a group of conditions characterized by abnormally high blood glucose levels. This form of diabetes usually begins before age 30. *GCK* gene mutations cause a type known as *GCK*-MODY (also called MODY2). Affected individuals usually have mildly elevated blood glucose levels from birth, although they typically have no symptoms associated with the condition, and diabetes-related complications are extremely rare.

Most *GCK* gene mutations involved in *GCK*-MODY change single protein building blocks in the glucokinase protein or result in an abnormally short version of the protein. The altered protein may be broken down, or the function may be impaired, reducing glucokinase activity in cells. As a result, beta cells are less able to detect changes in blood glucose and release insulin to control it, so blood glucose remains elevated.

Congenital hyperinsulinism

MedlinePlus Genetics provides information about Congenital hyperinsulinism

Gestational diabetes

MedlinePlus Genetics provides information about Gestational diabetes

Permanent neonatal diabetes mellitus

MedlinePlus Genetics provides information about Permanent neonatal diabetes mellitus

Other Names for This Gene

- ATP:D-glucose 6-phosphotransferase
- HEXOKINASE 4
- Hexokinase type IV
- HK4

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GCK (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2645[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GCK%5BTI%5D%29+OR+%28glucokinase%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GLUCOKINASE; GCK (https://omim.org/entry/138079)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2645)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GCK[gene])

References

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- Steele AM, Shields BM, Wensley KJ, Colclough K, Ellard S, Hattersley AT. Prevalence of vascular complications among patients with glucokinase mutationsand prolonged, mild hyperglycemia. JAMA. 2014 Jan 15;311(3):279-86. doi:10.1001/jama.2013.283980. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24430320)

Genomic Location

The *GCK* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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