GCK Gene

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Glucokinase

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1. 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 sugar 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.

2. Health Conditions Related to Genetic Changes

2.1 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 sugar 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 sugar 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 sugar and release insulin to control it, so blood sugar remains elevated.

2.2 Congenital Hyperinsulinism

2.3 Gestational Diabetes

2.4 Permanent Neonatal Diabetes Mellitus

3. Other Names for This Gene

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

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