# GLDC Gene

Subjects: Genetics & Heredity
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Glycine Decarboxylase

genes

## 1. Normal Function

The *GLDC* gene provides instructions for making an enzyme called glycine dehydrogenase. This protein is one of four enzymes that work together in a group called the glycine cleavage system. Within cells, this system is active in specialized energy-producing centers called mitochondria.

As its name suggests, the glycine cleavage system breaks down a molecule called glycine by cutting (cleaving) it into smaller pieces. Glycine is an amino acid, which is a building block of proteins. This molecule also acts as a neurotransmitter, which is a chemical messenger that transmits signals in the brain. The breakdown of excess glycine when it is no longer needed is necessary for the normal development and function of nerve cells in the brain.

The breakdown of glycine by the glycine cleavage system produces a molecule called a methyl group. This molecule is added to and used by a vitamin called folate. Folate is required for many functions in the cell and is important for brain development.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Nonketotic hyperglycinemia

Mutations in the *GLDC* gene account for about 80 percent of all cases of nonketotic hyperglycinemia. This condition is characterized by abnormally high levels of glycine in the body (hyperglycinemia). Affected individuals have serious neurological problems. The signs and symptoms of the condition vary in severity and can include severe breathing difficulties shortly after birth as well as weak muscle tone (hypotonia), seizures, and delayed development of milestones. More than 400 mutations have been identified in affected individuals. Many of these genetic changes alter single amino acids in glycine dehydrogenase. Other mutations insert or delete genetic material in the *GLDC* gene, or disrupt how genetic information from the gene is spliced together to make a blueprint for producing glycine dehydrogenase.

Some *GLDC* gene mutations lead to the production of a nonfunctional version of glycine dehydrogenase, while other mutations reduce but do not eliminate the enzyme's activity. When an altered version of this enzyme is incorporated into the glycine cleavage system, it impairs the system's ability to break down glycine. As a result, excess glycine can build up in the body's organs and tissues. In addition, the production of methyl groups for use by folate is reduced. It is unclear how these abnormalities contribute to the developmental disability, seizures, breathing difficulties, and other features characteristic of nonketotic hyperglycinemia.

## 3. Other Names for This Gene

- GCE
- GCSP
- GCSP_HUMAN
- glycine cleavage system protein P
- glycine decarboxylase P-protein
- glycine dehydrogenase (decarboxylating)
- glycine dehydrogenase (decarboxylating; glycine decarboxylase, glycine cleavage system protein P)
- NKH

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