# GSS gene

glutathione synthetase

## Normal Function

The *GSS* gene provides instructions for making an enzyme called glutathione synthetase. Glutathione synthetase participates in a process called the gamma-glutamyl cycle. The gamma-glutamyl cycle is a sequence of chemical reactions that takes place in most of the body's cells. These reactions are necessary for the production of glutathione, a small molecule made of three protein building blocks (amino acids). Glutathione protects cells from damage caused by unstable oxygen-containing molecules, which are byproducts of energy production. Glutathione is called an antioxidant because of its role in protecting cells from the damaging effects of these unstable molecules. Glutathione also helps process medications and cancer-causing compounds (carcinogens), and helps build DNA, proteins, and other important cellular components.

## Health Conditions Related to Genetic Changes

Glutathione synthetase deficiency

More than 30 mutations in the *GSS* gene have been identified in people with glutathione synthetase deficiency. Characteristic features of this condition include the abnormal destruction of red blood cells (hemolytic anemia), the release of large amounts of a compound called 5-oxoproline in the urine (5-oxoprolinuria), and elevated acidity in the blood and tissues (metabolic acidosis). Severely affected individuals may also have neurological problems.

Most of the *GSS* mutations involved in glutathione synthetase deficiency change single amino acids in glutathione synthetase. Other mutations disrupt how genetic information from the *GSS* gene is pieced together to make a blueprint for producing the enzyme. The altered glutathione synthetase enzyme may be unstable, shorter than usual, or the wrong shape. All of these changes reduce the activity of the enzyme and disrupt the gamma-glutamyl cycle, preventing adequate production of glutathione. Low levels of glutathione affect other chemical reactions in the body, leading to the overproduction of 5-oxoproline. Accumulation of this compound in red blood cells and other tissues causes hemolytic anemia and metabolic acidosis, and its release leads to 5-oxoprolinuria.

### Other Names for This Gene

- gamma-L-Glutamyl-L-cysteine:glycine ligase (ADP-forming)
- glutathione synthase
- GSH synthetase
- GSHB_HUMAN
- GSHS
- MGC14098

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GSS (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2937[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GSS+gene%5BTIAB%5D%29+OR+%28glutathione+synthetase%5BTIAB%5D%29%29+OR+%28GSH+synthetase%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GLUTATHIONE SYNTHETASE; GSS (https://omim.org/entry/601002)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2937)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GSS[gene])

## References


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## Genomic Location

The *GSS* gene is found on chromosome 20 (https://medlineplus.gov/genetics/chromosome/20/).

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