

## L2HGDH gene

L-2-hydroxyglutarate dehydrogenase

### Normal Function

The *L2HGDH* gene provides instructions for making an enzyme called L-2-hydroxyglutarate dehydrogenase. This enzyme is found in mitochondria, which are the energy-producing centers within cells. Within mitochondria, the enzyme participates in reactions that produce energy for cell activities. Specifically, L-2-hydroxyglutarate dehydrogenase converts a compound called L-2-hydroxyglutarate to another compound called 2-ketoglutarate. A series of additional enzymes further process 2-ketoglutarate to produce energy.

### Health Conditions Related to Genetic Changes

#### 2-hydroxyglutaric aciduria

Researchers have identified more than 70 mutations in the *L2HGDH* gene that cause a type of 2-hydroxyglutaric aciduria known as L-2-hydroxyglutaric aciduria (L-2-HGA). This condition has a variety of signs and symptoms that result from progressive damage to the brain beginning early in life.

Some *L2HGDH* gene mutations change single protein building blocks (amino acids) in the L-2-hydroxyglutarate dehydrogenase enzyme, which likely impairs its function. Other mutations insert or delete genetic material in the gene or lead to the production of an abnormally short, nonfunctional version of the enzyme. With a shortage of functional enzyme, L-2-hydroxyglutarate is not broken down but instead builds up in cells. At high levels, this compound can damage cells and lead to cell death. Brain cells appear to be the most vulnerable to the toxic effects of this compound, which may explain why the signs and symptoms of L-2-HGA primarily involve the brain.

### Other Names for This Gene

- 2-hydroxyglutarate dehydrogenase
- alpha-hydroxyglutarate oxidoreductase
- alpha-ketoglutarate reductase
- C14orf160
- duranin

- FLJ12618
- L-alpha-hydroxyglutarate dehydrogenase
- L2HGDH\_HUMAN

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of L2HGDH ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79944\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79944[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28L2HGDH%5BTIAB%5D%29+OR+%28L-2-hydroxyglutarate+dehydrogenase%5BTIAB%5D%29%29+OR+%28%28duranin%5BTIAB%5D%29+OR+%28L-2-HGDH%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29>)

### Catalog of Genes and Diseases from OMIM

- L-2-HYDROXYGLUTARATE DEHYDROGENASE; L2HGDH (<https://omim.org/entry/609584>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/79944>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=L2HGDH\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=L2HGDH[gene]))

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## Genomic Location

The *L2HGDH* gene is found on chromosome 14 (<https://medlineplus.gov/genetics/chromosome/14/>).

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