

## HGD gene

homogentisate 1,2-dioxygenase

### Normal Function

The *HGD* gene provides instructions for making an enzyme called homogentisate oxidase, which is active chiefly in the liver and kidneys. This enzyme participates in a step-wise process that breaks down two protein building blocks (amino acids), phenylalanine and tyrosine, when they are no longer needed or are present in excess. These two amino acids also play a role in making certain hormones, pigments, and brain chemicals called neurotransmitters.

Homogentisate oxidase is responsible for a specific step in the breakdown of phenylalanine and tyrosine. Previous steps convert the two amino acids into a molecule called homogentisic acid. Homogentisate oxidase adds two oxygen atoms to homogentisic acid, converting it to another molecule called maleylacetoacetate. Other enzymes break down maleylacetoacetate into smaller molecules that are later used for energy or to make other products that can be used by the body.

### Health Conditions Related to Genetic Changes

#### Alkaptonuria

More than 65 mutations in the *HGD* gene have been identified in people with alkaptonuria. Most of these mutations change single amino acids used to build the homogentisate oxidase enzyme. For example, a substitution of the amino acid valine for the amino acid methionine at protein position 368 (also written as Met368Val) is the most common *HGD* mutation in European populations.

Mutations in the *HGD* gene inactivate homogentisate oxidase by changing its structure. Without a functional version of this enzyme, phenylalanine and tyrosine are not broken down properly and homogentisic acid builds up in the body. Excess homogentisic acid and related compounds are deposited in connective tissues such as cartilage and skin, which causes them to darken. Over time, a buildup of this substance in the joints leads to arthritis. Homogentisic acid is also excreted in urine, making the urine turn dark when exposed to air.

## Other Names for This Gene

- AKU
- HGD\_HUMAN
- HGO
- homogentisate 1,2-dioxygenase (homogentisate oxidase)
- Homogentisic acid oxidase
- homogentisicase

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of HGD ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3081\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3081[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28homogentisate+1,2-dioxygenase%5BTIAB%5D%29+OR+%28%28homogentisate+1,2-dioxygenase%5BTIAB%5D%29+OR+%28homogentisicase%5BTIAB%5D%29+OR+%28Homogentisic+acid+oxidase%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- HOMOGENITISATE 1,2-DIOXYGENASE; HGD (<https://omim.org/entry/607474>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3081>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=HGD\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=HGD[gene]))

## References

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## Genomic Location

The *HGD* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

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