DHODH gene

dihydroorotate dehydrogenase (quinone)

Normal Function

The *DHODH* gene provides instructions for making an enzyme called dihydroorotate dehydrogenase. This enzyme is involved in producing pyrimidines, which are building blocks of DNA, its chemical cousin RNA, and molecules such as ATP and GTP that serve as energy sources in the cell. Dihydroorotate dehydrogenase functions within mitochondria, the energy-producing centers within cells. Specifically, this enzyme converts a molecule called dihydroorotate to a molecule called orotic acid. In subsequent steps, other enzymes modify orotic acid to produce pyrimidines.

Health Conditions Related to Genetic Changes

Miller syndrome

At least 11 mutations in the *DHODH* gene have been found to cause Miller syndrome. Most of these mutations change single protein building blocks (amino acids) in dihydroorotate dehydrogenase, which likely impairs the enzyme's ability to function normally. It is unclear exactly how *DHODH* gene mutations lead to the signs and symptoms of Miller syndrome.

Other Names for This Gene

- DHOdehase
- dihydroorotate dehydrogenase
- dihydroorotate dehydrogenase, mitochondrial
- dihydroorotate dehydrogenase, mitochondrial precursor
- dihydroorotate oxidase
- human complement of yeast URA1
- POADS
- PYRD_HUMAN
- URA1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DHODH (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1723[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DHODH%5BTIAB%5D%29+OR+%28dihydroorotate+dehydrogenase%5BTIAB%5D%29%29+OR+%28%28POADS%5BTIAB%5D%29+OR+%28DHOdehase%5BTIAB%5D%29+OR+%28dihydroorotate+oxidase%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+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- DIHYDROOROTATE DEHYDROGENASE; DHODH (https://omim.org/entry/126064)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1723)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DHODH[gene])

References

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Genomic Location

The *DHODH* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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