



QDPR gene

quinoid dihydropteridine reductase

Normal Function

The *QDPR* gene provides instructions for making an enzyme called quinoid dihydropteridine reductase. This enzyme helps carry out one step in the chemical pathway that recycles a molecule called tetrahydrobiopterin (BH₄).

Tetrahydrobiopterin plays a critical role in processing several protein building blocks (amino acids) in the body. For example, it works with the enzyme phenylalanine hydroxylase to convert an amino acid called phenylalanine into another amino acid, tyrosine. Tetrahydrobiopterin is also involved in reactions that produce chemicals called neurotransmitters, which transmit signals between nerve cells in the brain. Because it helps enzymes carry out chemical reactions, tetrahydrobiopterin is known as a cofactor.

When tetrahydrobiopterin interacts with enzymes during chemical reactions, the cofactor is altered and must be recycled to a usable form. Quinoid dihydropteridine reductase is one of two enzymes that help recycle tetrahydrobiopterin in the body.

Health Conditions Related to Genetic Changes

Tetrahydrobiopterin deficiency

More than 30 mutations in the *QDPR* gene have been found to cause tetrahydrobiopterin deficiency. When this condition results from *QDPR* gene mutations, it is known as dihydropteridine reductase (DHPR) deficiency. DHPR deficiency accounts for about one-third of all cases of tetrahydrobiopterin deficiency.

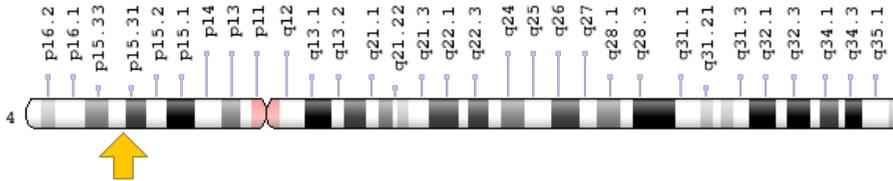
Most *QDPR* gene mutations change single amino acids in quinoid dihydropteridine reductase, although some mutations insert small amounts of DNA into the *QDPR* gene or disrupt the way the gene's instructions are used to make the enzyme. Changes in quinoid dihydropteridine reductase greatly reduce or eliminate the enzyme's activity. Without enough of this enzyme, tetrahydrobiopterin is not recycled properly. As a result, this cofactor is not available to participate in chemical reactions such as the conversion of phenylalanine to tyrosine. If phenylalanine is not converted to tyrosine, it can build up to toxic levels in the blood and other tissues. Nerve cells in the brain are particularly sensitive to phenylalanine levels, which is why excessive amounts of this substance can cause brain damage.

Additionally, a reduction in quinoid dihydropteridine reductase activity disrupts the production of certain neurotransmitters in the brain. Because neurotransmitters are necessary for normal brain function, changes in the levels of these brain chemicals contribute to intellectual disability in people with DHPR deficiency.

Chromosomal Location

Cytogenetic Location: 4p15.32, which is the short (p) arm of chromosome 4 at position 15.32

Molecular Location: base pairs 17,486,393 to 17,512,234 on chromosome 4 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DHPR
- DHPR_HUMAN
- Dihydropteridine reductase
- PKU2
- SDR33C1

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28QDPR%5BTIAB%5D%29+OR+%28quinoid+dihydropteridine+reductase%5BTIAB%5D%29%29+OR+%28Dihydropteridine+reductase%5BTIAB%5D%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

- QUINOID DIHYDROPTERIDINE REDUCTASE
<http://omim.org/entry/612676>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=QDPR%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:9752
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:5860>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/5860>
- UniProt
<https://www.uniprot.org/uniprot/P09417>

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