

PPOX gene

protoporphyrinogen oxidase

Normal Function

The *PPOX* gene provides instructions for making an enzyme known as protoporphyrinogen oxidase. This enzyme is involved in the production of a molecule called heme. Heme is vital for all of the body's organs, although it is most abundant in the blood, bone marrow, and liver. Heme is an essential component of iron-containing proteins called hemoproteins, including hemoglobin (the protein that carries oxygen in the blood).

The production of heme is a multi-step process that requires eight different enzymes. Protoporphyrinogen oxidase is responsible for the seventh step in this process, in which two hydrogen atoms are removed from protoporphyrinogen IX (the product of the sixth step) to form protoporphyrin IX. In the final step, another enzyme modifies protoporphyrin IX by inserting an iron atom to produce heme.

Health Conditions Related to Genetic Changes

Porphyria

More than 130 mutations in the *PPOX* gene have been identified in people with a form of porphyria called variegate porphyria. A particular *PPOX* gene mutation is found in about 95 percent of South African families with this form of the disorder. The mutation changes a single protein building block (amino acid) in protoporphyrinogen oxidase, replacing the amino acid arginine with the amino acid tryptophan at position 59 (written as Arg59Trp or R59W).

Mutations in the *PPOX* gene reduce the activity of protoporphyrinogen oxidase, allowing compounds called porphyrin precursors to build up in the body. These compounds are formed during the normal process of heme production, but reduced activity of protoporphyrinogen oxidase allows them to accumulate to toxic levels. Nongenetic factors such as certain drugs, alcohol, dieting, as well as other genetic factors that have not been identified, also contribute to the characteristic features of variegate porphyria.

Other Names for This Gene

- PPO

- PPOX_HUMAN
- protoporphyrinogen dehydrogenase
- protoporphyrinogen IX oxidase
- protoporphyrinogenase
- protox

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PPOX ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5498\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5498[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PPOX%5BTIAB%5D%29+OR+%28protoporphyrinogen+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+1800+days%22%5Bdp%5D%29%29>)

Catalog of Genes and Diseases from OMIM

- PROTOPORPHYRINOGEN OXIDASE; PPOX (<https://omim.org/entry/600923>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5498>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PPOX\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PPOX[gene]))

References

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Genomic Location

The *PPOX* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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