

CPOX gene

coproporphyrinogen oxidase

Normal Function

The *CPOX* gene provides instructions for making an enzyme known as coproporphyrinogen 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. Coproporphyrinogen oxidase is responsible for the sixth step in this process, the removal of carbon and oxygen atoms from coproporphyrinogen III (the product of the fifth step) to form protoporphyrinogen IX. In subsequent steps, two other enzymes modify protoporphyrinogen IX and incorporate an iron atom to produce heme.

Health Conditions Related to Genetic Changes

Porphyria

At least 45 mutations in the *CPOX* gene have been found to cause porphyria. Mutations in this gene can cause two types of porphyria: hereditary coproporphyria and a variant known as harderoporphyria.

Most *CPOX* gene mutations change single protein building blocks (amino acids) in coproporphyrinogen oxidase. A single mutation appears to be responsible for harderoporphyria; this genetic change replaces the amino acid glycine with the amino acid glutamic acid at position 404 (written as Lys404Glu or K404E). Mutations in the *CPOX* gene reduce the activity of coproporphyrinogen oxidase, allowing compounds called porphyrins to build up in the body. These compounds are formed during the normal process of heme production, but reduced activity of coproporphyrinogen oxidase allows them to accumulate to toxic levels. This buildup, in combination with nongenetic factors such as certain drugs, alcohol, and dieting, leads to the signs and symptoms of hereditary coproporphyria and harderoporphyria.

Other Names for This Gene

- Coprogen oxidase
- Coproporphyrinogen III oxidase, mitochondrial
- Coproporphyrinogen III Oxidases
- coproporphyrinogen oxidase (coproporphyria, harderopporphyria)
- Coproporphyrinogen:oxygen oxidoreductase (decarboxylating)
- Coproporphyrinogenase
- COX
- CPO
- CPX
- HCP
- HEM6_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CPOX ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1371\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1371[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CPOX+gene%5BTIAB%5D%29+OR+%28coproporphyrinogen+oxidase%5BTIAB%5D%29+OR+%28Coproporphyrinogen+III+Oxidase%5BTIAB%5D%29+OR+%28Coproporphyrinogen%5BTIAB%5D%29+OR+%28CPO+gene%5BTIAB%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

- COPROPORPHYRINOGEN OXIDASE; CPOX (<https://omim.org/entry/612732>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1371>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CPOX\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CPOX[gene]))

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Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15868463>)

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Genomic Location

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

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