

UROS gene

uroporphyrinogen III synthase

Normal Function

The *UROS* gene provides instructions for making an enzyme known as uroporphyrinogen III synthase. 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. Uroporphyrinogen III synthase is responsible for the fourth step in this process, in which hydroxymethylbilane (the product of the third step) is rearranged to form uroporphyrinogen III. In subsequent steps, four other enzymes produce and modify compounds that ultimately lead to heme.

Health Conditions Related to Genetic Changes

Porphyria

More than 35 mutations in the *UROS* gene have been found to cause a form of porphyria known as congenital erythropoietic porphyria. Most of these mutations change single protein building blocks (amino acids) in uroporphyrinogen III synthase. The most common *UROS* gene mutation, which is found in about one-third of all cases of congenital erythropoietic porphyria, replaces the amino acid cysteine with the amino acid arginine at position 73 (written as Cys73Arg or C73R). Several other mutations occur in a nearby region of DNA that regulates the activity of the *UROS* gene.

Mutations in or near the *UROS* gene alter the structure and function of uroporphyrinogen III synthase, which reduces the enzyme's activity. A shortage of functional uroporphyrinogen III synthase allows compounds called porphyrins to build up in developing red blood cells. These compounds are formed during the normal process of heme production, but reduced activity of uroporphyrinogen III synthase allows them to accumulate to toxic levels. The excess porphyrins can leak out of developing red blood cells and be transported through the bloodstream to the skin and other tissues. An accumulation of these substances in the skin causes oversensitivity to sunlight and the other characteristic features of congenital erythropoietic porphyria.

Other Names for This Gene

- Cosynthase
- HEM4_HUMAN
- Hydroxymethylbilane hydro-lyase (cyclizing)
- UROIIIS
- Uroporphyrinogen co-synthetase
- Uroporphyrinogen III Cosynthetase
- uroporphyrinogen III synthase (congenital erythropoietic porphyria)
- Uroporphyrinogen III Synthetase
- Uroporphyrinogen Isomerase
- Uroporphyrinogen-III cosynthase
- Uroporphyrinogen-III Synthase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of UROS ([https://www.ncbi.nlm.nih.gov/qtr/all/tests/?term=7390\[geneid\]](https://www.ncbi.nlm.nih.gov/qtr/all/tests/?term=7390[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28UROS%5BTIAB%5D%29+OR+%28uroporphyrinogen+III+synthase%5BTIAB%5D%29%29+OR+%28%28UROIIIS%5BTIAB%5D%29+OR+%28Uroporphyrinogen+III+Cosynthetase%5BTIAB%5D%29+OR+%28Uroporphyrinogen+III+Synthetase%5BTIAB%5D%29+OR+%28Uroporphyrinogen+Isomerase%5BTIAB%5D%29+OR+%28Uroporphyrinogen-III+cosynthase%5BTIAB%5D%29+OR+%28Uroporphyrinogen-III+Synthase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+3240+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- UROPORPHYRINOGEN III SYNTHASE; UROS (<https://omim.org/entry/606938>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7390>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=UROS\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=UROS[gene]))

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Genomic Location

The *UROS* gene is found on chromosome 10 (<https://medlineplus.gov/genetics/chromosome/10/>).

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