# POR gene

cytochrome p450 oxidoreductase

## Normal Function

The *POR* gene provides instructions for making the enzyme cytochrome P450 oxidoreductase. This enzyme is required for the normal functioning of more than 50 enzymes in the cytochrome P450 family. Cytochrome P450 enzymes are involved in the formation (synthesis) and breakdown (metabolism) of various molecules and chemicals within cells.

Cytochrome P450 enzymes are critical for the synthesis of cholesterol and steroid hormones. Cholesterol is a substance that has many essential functions both before and after birth, including roles in the production of steroid hormones and in the formation and growth of bones. Steroid hormones are needed for normal development and reproduction. This group of hormones includes testosterone and estrogen, which are essential for normal sexual development and reproduction; corticosteroids, which are involved in the body's response to stress; and aldosterone, which helps regulate the body's salt and water balance.

Additionally, cytochrome P450 enzymes are involved in the metabolism of ingested substances, such as medications, in the liver. Because cytochrome P450 oxidoreductase helps regulate the activity of these enzymes, researchers suspect that normal variations in the *POR* gene may influence a person's response to particular drugs (drug metabolism).

## Health Conditions Related to Genetic Changes

Cytochrome P450 oxidoreductase deficiency

More than 50 mutations in the *POR* gene have been found to cause cytochrome P450 oxidoreductase deficiency. This condition causes hormonal changes that can affect the development of the reproductive system, skeleton, and other parts of the body. The disorder affects sexual development before birth and at puberty, and severe cases are also characterized by skeletal abnormalities.

Most of the mutations that cause cytochrome P450 oxidoreductase deficiency change single protein building blocks (amino acids) in cytochrome P450 oxidoreductase. *POR* gene mutations significantly reduce the enzyme's activity, which disrupts the production

of steroid hormones. Changes in sex hormones such as testosterone and estrogen lead to problems with sexual development.

Reduced activity of cytochrome P450 oxidoreductase can also disrupt the production of cholesterol, which likely impairs normal bone formation in severe cases of cytochrome P450 oxidoreductase deficiency. Studies suggest that a molecule called retinoic acid also plays a role in the skeletal abnormalities found in severe cases. The breakdown of retinoic acid requires cytochrome P450 oxidoreductase; if a shortage of cytochrome P450 oxidoreductase prevents retinoic acid from being broken down, the resulting excess of that molecule can stimulate the abnormal growth and fusion of bones.

It is unclear whether mutations in the *POR* gene affect how the liver processes medications. The role of this enzyme in drug metabolism is an active area of research.

## Other Names for This Gene

- CPR
- CYPOR
- cytochrome P450 reductase
- FLJ26468
- NADPH-dependent cytochrome P450 reductase
- NCPR_HUMAN
- P450 (cytochrome) oxidoreductase
- P450R

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of POR (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5447[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28P450+oxidoreductase%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+1440+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CYTOCHROME P450 OXIDOREDUCTASE; POR (https://omim.org/entry/124015)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5447)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=POR[gene])

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## Genomic Location

The *POR* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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