CYP27B1 gene

cytochrome P450 family 27 subfamily B member 1

Normal Function

The *CYP27B1* gene provides instructions for making an enzyme called 1-alpha-hydroxylase (1α-hydroxylase). This enzyme carries out the second of two reactions to convert vitamin D to its active form, 1,25-dihydroxyvitamin D_3, also known as calcitriol. Vitamin D can be acquired from foods in the diet or can be made in the body with the help of sunlight exposure. When active, this vitamin is involved in maintaining the proper balance of several minerals in the body, including calcium and phosphate, which are essential for the normal formation of bones and teeth. One of vitamin D's major roles is to control the absorption of calcium and phosphate from the intestines into the bloodstream. Vitamin D is also involved in several processes unrelated to bone and tooth formation.

Health Conditions Related to Genetic Changes

Vitamin D-dependent rickets

At least 70 mutations in the *CYP27B1* gene have been found to cause vitamin D-dependent rickets type 1A (VDDR1A), also known as vitamin D 1α-hydroxylase deficiency. This disorder of bone development is characterized by low levels of calcium (hypocalcemia) and phosphate (hypophosphatemia) in the blood, which lead to soft, weak bones that are prone to fracture. A common feature of this condition is abnormally curved (bowed) legs.

The *CYP27B1* gene mutations that cause this condition reduce or eliminate the function of 1α-hydroxylase. As a result, vitamin D does not get converted to its active form and cannot control mineral absorption. The resulting reduction in calcium and phosphate absorption from the intestines into the blood means there is less of these minerals to be deposited in developing bones (bone mineralization), which leads to soft, weak bones and other features of VDDR1A. Hypocalcemia also causes muscle weakness and seizures in some affected individuals.

Autoimmune Addison disease

MedlinePlus Genetics provides information about Autoimmune Addison disease

Multiple sclerosis

MedlinePlus Genetics provides information about Multiple sclerosis

Other Names for This Gene

- 1alpha(OH)ase
- 25 hydroxyvitamin D3-1-alpha hydroxylase
- 25-hydroxyvitamin D-1 alpha hydroxylase, mitochondrial
- 25-OHD-1 alpha-hydroxylase
- CYP1alpha
- CYP27B
- cytochrome p450 27B1
- cytochrome P450 subfamily XXVIIB polypeptide 1
- cytochrome P450, family 27, subfamily B, polypeptide 1
- cytochrome P450C1 alpha
- cytochrome P450VD1-alpha
- P450c1
- VD3 1A hydroxylase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CYP27B1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1594[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28CYP27B1%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CYTOCHROME P450, SUBFAMILY XXVIIB, POLYPEPTIDE 1; CYP27B1 (https://omim.org/entry/609506)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1594)

- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CYP27B1[gene])

References

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- Ryan JW, Anderson PH, Turner AG, Morris HA. Vitamin D activities and metabolicbone disease. Clin Chim Acta. 2013 Oct 21;425:148-52. doi:10.1016/j.cca.2013.07.024. Epub 2013 Jul 30. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23911750)

Genomic Location

The *CYP27B1* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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