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.

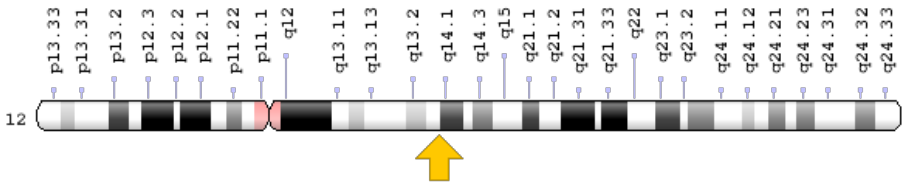
Autoimmune Addison disease

Multiple sclerosis

Chromosomal Location

Cytogenetic Location: 12q14.1, which is the long (q) arm of chromosome 12 at position 14.1

Molecular Location: base pairs 57,762,334 to 57,767,078 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 1alpha(OH)ase
- 25-hydroxyvitamin D-1 alpha hydroxylase, mitochondrial
- 25 hydroxyvitamin D3-1-alpha hydroxylase
- 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

Educational Resources

- Dietary Reference Intakes for Calcium, Phosphorus, Magnesium, Vitamin D, and Fluoride (1997): Vitamin D
  https://www.ncbi.nlm.nih.gov/books/NBK109831/
- Endocrinology: An Integrated Approach (2001): Classical Actions of Vitamin D on Intestine and Bone
  https://www.ncbi.nlm.nih.gov/books/NBK24/#_A788_

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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
  http://omim.org/entry/609506

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CYP27B1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CYP27B1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2606
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1594
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1594
- UniProt
  https://www.uniprot.org/uniprot/O15528

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