CYP2R1 gene

cytochrome P450 family 2 subfamily R member 1

Normal Function

The *CYP2R1* gene provides instructions for making an enzyme called 25-hydroxylase. This enzyme carries out the first of two reactions to convert vitamin D to its active form, 1,25-dihydroxyvitamin D3, 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 four mutations in the *CYP2R1* gene have been found to cause vitamin D-dependent rickets type 1B (VDDR1B). 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 *CYP2R1* gene mutations that cause this condition severely reduce or eliminate the function of 25-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 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 VDDR1B. Hypocalcemia also causes muscle weakness in some affected individuals.

Other Names for This Gene

- cytochrome P450 2R1
- cytochrome P450, family 2, R1
- cytochrome P450, family 2, subfamily R, polypeptide 1

- vitamin D 25-hydroxylase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CYP2R1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=120227[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CYP2R1%5BTIAB%5D%29+OR+%28cytochrome+P450+family+2+subfamily+R+member+1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CYTOCHROME P450, SUBFAMILY IIR, POLYPEPTIDE 1; CYP2R1 (https://omim.org/entry/608713)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/120227)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CYP2R1[gene])

References

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Genomic Location

The *CYP2R1* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

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