VDR gene

vitamin D receptor

Normal Function

The *VDR* gene provides instructions for making a protein called vitamin D receptor (VDR), which allows the body to respond to vitamin D. This vitamin can be acquired from foods in the diet or made in the body with help from sunlight exposure. Vitamin D 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.

The VDR protein attaches (binds) to the active form of vitamin D, known as calcitriol. This interaction allows VDR to partner with another protein called retinoid X receptor (RXR). The resulting complex then binds to particular regions of DNA, known as vitamin D response elements, and regulates the activity of vitamin D-responsive genes. By turning these genes on or off, the complex helps control calcium and phosphate absorption and other processes.

Although the mechanism is not completely understood, the VDR protein is also involved in hair growth. Studies suggest that this process does not require calcitriol binding.

Health Conditions Related to Genetic Changes

Vitamin D-dependent rickets

Mutations in the *VDR* gene cause vitamin D-dependent rickets type 2A (VDDR2A), also known as hereditary vitamin D-resistant rickets (HVDRR). 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 (rickets) that are prone to fracture. A common feature of this condition is abnormally curved (bowed) legs.

The *VDR* gene mutations that cause this condition prevent the VDR protein from functioning properly. Some changes in the *VDR* gene lead to an abnormally short version of the VDR protein. Others result in the production of an abnormal receptor that cannot bind to calcitriol, to RXR, or to DNA. Although there is plenty of calcitriol in the body, the altered VDR protein cannot stimulate gene activity important for mineral

absorption. The lack of calcium and phosphate absorption in the intestines slows deposition of these minerals into developing bone (bone mineralization), which leads to soft, weak bones and other features of VDDR2A. Hypocalcemia also causes muscle weakness and seizures in some affected individuals. Most *VDR* gene mutations impair hair growth, leading to hair loss (alopecia); however, mutations that block VDR's ability to interact with calcitriol do not cause alopecia, indicating that calcitriol is not necessary for the receptor's role in hair development.

Alopecia areata

MedlinePlus Genetics provides information about Alopecia areata

Intervertebral disc disease

MedlinePlus Genetics provides information about Intervertebral disc disease

Kidney stones

MedlinePlus Genetics provides information about Kidney stones

Leprosy

MedlinePlus Genetics provides information about Leprosy

Other Names for This Gene

- 1,25-dihydroxyvitamin D3 receptor
- NR1I1
- nuclear receptor subfamily 1 group I member 1
- vitamin D (1,25- dihydroxyvitamin D3) receptor
- vitamin D3 receptor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of VDR (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7421[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28VDR%5BTI%5D%29+OR+%28vitamin+D+++receptor%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- VITAMIN D RECEPTOR; VDR (https://omim.org/entry/601769)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7421)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=VDR[gene])

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Genomic Location

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

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