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 appropriately to vitamin D. This vitamin can be acquired from foods in the diet or made in the body with help from sunlight. 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 process unrelated to bone formation.

VDR 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 of proteins 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, VDR helps control calcium and phosphate absorption and other processes.

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

Health Conditions Related to Genetic Changes

intervertebral disc disease

vitamin D-dependent rickets

Mutations in the *VDR* gene cause vitamin D-dependent rickets type 2 (VDDR2), 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 bowed legs.

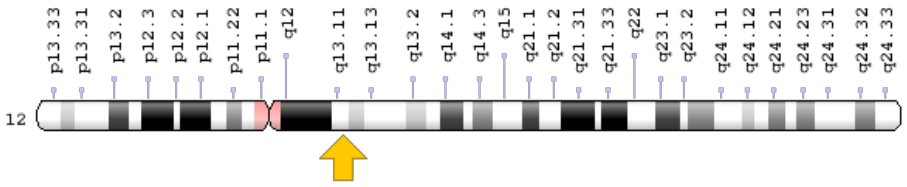
The *VDR* gene mutations that cause this condition prevent the VDR protein from functioning properly. Some changes in the *VDR* gene lead to an abnormally short version of the VDR protein; others result in the production of an abnormal receptor that cannot bind to calcitriol, to RXR, or to DNA. Despite plenty of calcitriol in the body, the altered VDR 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 VDDR2. Hypocalcemia also causes muscle weakness and seizures in some affected individuals. Most *VDR* gene mutations impair hair growth, leading to 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.

Chromosomal Location

Cytogenetic Location: 12q13.11, which is the long (q) arm of chromosome 12 at position 13.11

Molecular Location: base pairs 47,841,537 to 47,905,031 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

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

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_
- Molecular Cell Biology (fourth edition, 2000): Lipid-Soluble Hormones Control the Activities of Nuclear Receptors
  https://www.ncbi.nlm.nih.gov/books/NBK21677/#_A2652_

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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

OMIM

- VITAMIN D RECEPTOR
  http://omim.org/entry/601769

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_VDR.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=VDR%5Bgene%5D
- HGNC Gene Family: Nuclear hormone receptors
  http://www.genenames.org/cgi-bin/genefamilies/set/71
- HGNC Gene Family: Protein phosphatase 1 regulatory subunits
  http://www.genenames.org/cgi-bin/genefamilies/set/694
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12679

- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7421
- UniProt
  http://www.uniprot.org/uniprot/P11473

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