CYP4V2 gene

cytochrome P450 family 4 subfamily V member 2

Normal Function

The *CYP4V2* gene provides instructions for making a member of the cytochrome P450 family of enzymes. These enzymes are involved in the formation and breakdown of various molecules and chemicals within cells. The CYP4V2 enzyme is involved in a multi-step process called fatty acid oxidation in which fats are broken down and converted into energy, but the enzyme's specific function is not well understood.

Health Conditions Related to Genetic Changes

Bietti crystalline dystrophy

At least 42 *CYP4V2* gene mutations have been identified in people with Bietti crystalline dystrophy, a disorder in which numerous small, yellow or white crystal-like deposits of fatty (lipid) compounds accumulate in the light-sensitive tissue that lines the back of the eye (the retina). The deposits damage the retina, resulting in progressive vision loss.

CYP4V2 gene mutations that cause Bietti crystalline dystrophy are predicted to change the structure of the CYP4V2 enzyme in a way that reduces or eliminates its activity. The mutations likely affect lipid breakdown; however, it is unknown how they lead to the specific signs and symptoms of Bietti crystalline dystrophy. For unknown reasons, the severity of the signs and symptoms differs significantly among individuals with the same *CYP4V2* gene mutation.

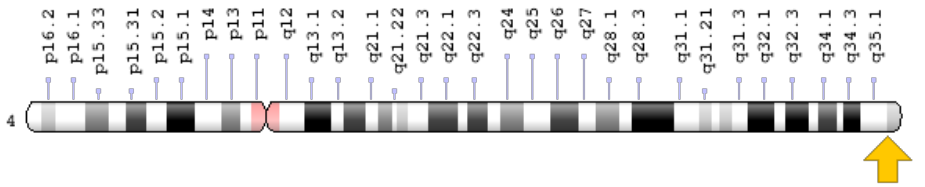
Other disorders

Certain common variations (polymorphisms) in the *CYP4V2* gene have been associated with an increased risk of a type of blood clot called a deep venous thrombosis (DVT). DVTs occur most often in the deep veins of the legs or arms. If these clots travel through the bloodstream, they can lodge in the lungs and cause a life-threatening complication called a pulmonary embolism. While nearby genes on chromosome 4 are known to be involved in blood clotting, it is unclear how the *CYP4V2* gene variations may affect this process.

Chromosomal Location

Cytogenetic Location: 4q35.1-q35.2, which is the long (q) arm of chromosome 4 between positions 35.1 and 35.2

Molecular Location: base pairs 186,191,567 to 186,213,463 on chromosome 4 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BCD
- CP4V2_HUMAN
- CYP4AH1
- cytochrome P450 4V2
- cytochrome P450, family 4, subfamily V, polypeptide 2

Additional Information & Resources

Clinical Information from GeneReviews

- Bietti Crystalline Dystrophy
  https://www.ncbi.nlm.nih.gov/books/NBK91457

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28CYP4V2%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+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- CYTOCHROME P450, FAMILY 4, SUBFAMILY V, POLYPEPTIDE 2
  http://omim.org/entry/608614

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CYP4V2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:23198
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:285440
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/285440
- UniProt
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