DPYD gene

dihydropyrimidine dehydrogenase

Normal Function

The *DPYD* gene provides instructions for making an enzyme called dihydropyrimidine dehydrogenase, which is involved in the breakdown of molecules called uracil and thymine when they are not needed. Uracil and thymine are pyrimidines, which are one type of nucleotide. Nucleotides are building blocks of DNA, its chemical cousin RNA, and molecules such as ATP and GTP that serve as energy sources in the cell.

Dihydropyrimidine dehydrogenase is involved in the first step of the breakdown of pyrimidines. This enzyme converts uracil to another molecule called 5,6-dihydrouracil and converts thymine to 5,6-dihydrothymine. The molecules created when pyrimidines are broken down are excreted by the body or used in other cellular processes.

Health Conditions Related to Genetic Changes

Dihydropyrimidine dehydrogenase deficiency

More than 50 mutations in the *DPYD* gene have been identified in people with dihydropyrimidine dehydrogenase deficiency. *DPYD* gene mutations interfere with the breakdown of uracil and thymine and result in excess quantities of these molecules in the blood, urine, and the fluid that surrounds the brain and spinal cord (cerebrospinal fluid). It is unclear how the excess uracil and thymine are related to the specific neurological problems that affect some people with dihydropyrimidine dehydrogenase deficiency.

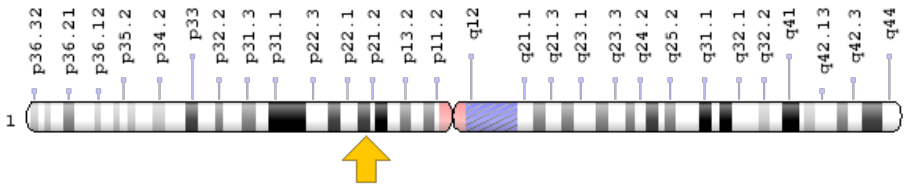
Mutations in the *DPYD* gene also interfere with the breakdown of drugs with structures similar to the pyrimidines, such as the cancer drugs 5-fluorouracil and capecitabine. As a result, these drugs accumulate in the body and cause the severe reactions that can occur in people with dihydropyrimidine dehydrogenase deficiency.

Coloboma

Chromosomal Location

Cytogenetic Location: 1p21.3, which is the short (p) arm of chromosome 1 at position 21.3

Molecular Location: base pairs 97,077,743 to 97,921,059 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DHP
- DHPDHASE
- dihydropyrimidine dehydrogenase [NADP+]
- dihydrothymine dehydrogenase
- dihydrouracil dehydrogenase
- DPD
- DPYD_HUMAN
- MGC132008
- MGC70799

Additional Information & Resources

Educational Resources

- Holland-Frei Cancer Medicine (sixth edition, 2003): Pyrimidine Analogs: Catabolic Reactions
  https://www.ncbi.nlm.nih.gov/books/NBK13287/#A12024

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DPYD%5BTIAB%5D%29+OR+%28dihydropyrimidine+dehydrogenase%5BTIAB%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+720+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- DIHYDROPYRIMIDINE DEHYDROGENASE
  http://omim.org/entry/612779

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DPYD.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DPYD%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3012
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1806
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1806
- UniProt
  https://www.uniprot.org/uniprot/Q12882

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