CYP27A1 gene

cytochrome P450 family 27 subfamily A member 1

Normal Function

The *CYP27A1* gene is a member of the cytochrome P450 gene family. Enzymes produced from the cytochrome P450 genes are involved in the formation and breakdown of various molecules and chemicals within cells. The *CYP27A1* gene provides instructions for producing an enzyme called sterol 27-hydroxylase. This enzyme is located in the energy-producing centers of cells (mitochondria), where it is involved in the pathway that breaks down cholesterol to form acids used to digest fats (bile acids). Specifically, sterol 27-hydroxylase breaks down cholesterol to form a bile acid called chenodeoxycholic acid. The formation of bile acids from cholesterol is the body's main pathway for cholesterol removal. Sterol 27-hydroxylase plays a key role in maintaining normal cholesterol levels in the body.

Health Conditions Related to Genetic Changes

Cerebrotendinous xanthomatosis

At least 90 mutations that cause cerebrotendinous xanthomatosis have been identified in the *CYP27A1* gene. Cerebrotendinous xanthomatosis is a disorder characterized by abnormal storage of fats (lipids) in many areas of the body. Most *CYP27A1* gene mutations change one protein building block (amino acid) in the sterol 27-hydroxylase enzyme. The most common mutation changes the amino acid arginine to the amino acid cysteine at position 362 in the protein (written as Arg362Cys or R362C). Changes in amino acids typically disrupt the normal function of the protein and impair its ability to help form chenodeoxycholic acid. Other mutations cause no functional enzyme to be made. As a result, other molecules are formed by an alternative pathway. A molecule called cholestanol, which is similar to cholesterol, is produced and accumulates in blood and tissues. Cholesterol also accumulates in tissues, but levels in blood are typically normal. The accumulation of cholesterol and cholestanol throughout the body's tissues causes the signs and symptoms of cerebrotendinous xanthomatosis.
Chromosomal Location

Cytogenetic Location: 2q35, which is the long (q) arm of chromosome 2 at position 35
Molecular Location: base pairs 218,781,733 to 218,815,293 on chromosome 2 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

• 5-beta-cholestane-3-alpha, 7-alpha, 12-alpha-triol 27-hydroxylase
• CP27
• CP27A_HUMAN
• CTX
• CYP27
• cytochrome P-450C27/25
• cytochrome P450, family 27, subfamily A, polypeptide 1
• cytochrome P450, subfamily XXVIIA (steroid 27-hydroxylase, cerebrotendinous xanthomatosis), polypeptide 1
• sterol 27-hydroxylase
• vitamin D(3) 25-hydroxylase

Additional Information & Resources

Educational Resources

• Biochemistry (fifth edition, 2002): Important Derivatives of Cholesterol Include Bile Salts and Steroid Hormones
  https://www.ncbi.nlm.nih.gov/books/NBK22339/

Clinical Information from GeneReviews

• Cerebrotendinous Xanthomatosis
  https://www.ncbi.nlm.nih.gov/books/NBK1409
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28CYP27A1%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+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- CYTOCHROME P450, SUBFAMILY XXVIIA, POLYPEPTIDE 1
  http://omim.org/entry/606530

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
- ClinVar
- HGNC Gene Family: Cytochrome P450 family 27
  https://www.genenames.org/cgi-bin/genefamilies/set/1014
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1593
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q02318

Sources for This Summary

- OMIM: CYTOCHROME P450, SUBFAMILY XXVIIA, POLYPEPTIDE 1
  http://omim.org/entry/606530
Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16816916

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/25424010 
Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4264335/

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12777473

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12117727

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