GNPAT gene

glyceronephosphate O-acyltransferase

Normal Function

The *GNPAT* gene provides instructions for making an enzyme known as glyceronephosphate O-acyltransferase (GNPAT) or dihydroxyacetonephosphate acyltransferase (DHAPAT). This enzyme is found in structures called peroxisomes, which are sac-like compartments within cells that contain enzymes needed to break down many different substances. Peroxisomes are also important for the production of fats (lipids) used in digestion and in the nervous system.

Within peroxisomes, the DHAPAT enzyme is responsible for the first step in the production of lipid molecules called plasmalogens. These molecules are found in cell membranes throughout the body. They are also abundant in myelin, which is the protective substance that covers nerve cells. However, little is known about the functions of plasmalogens. Researchers suspect that these molecules may help protect cells from oxidative stress, which occurs when unstable molecules called free radicals accumulate to levels that damage or kill cells. Plasmalogens may also play important roles in interactions between lipids and proteins, the transmission of chemical signals in cells, and the fusion of cell membranes.

Health Conditions Related to Genetic Changes

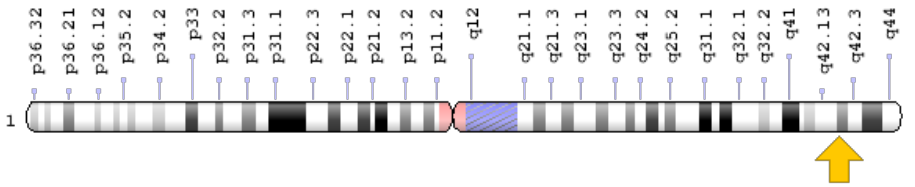
Rhizomelic chondrodysplasia punctata

At least five mutations in the *GNPAT* gene have been found to cause rhizomelic chondrodysplasia punctata type 2 (RCDP2). These mutations prevent cells from making any functional DHAPAT enzyme. A shortage of this enzyme disrupts peroxisome function and severely reduces the amount of plasmalogens within cells. It is unclear how these abnormalities lead to shortened long bones, intellectual disability, and the other characteristic features of RCDP2.

Chromosomal Location

Cytogenetic Location: 1q42.2, which is the long (q) arm of chromosome 1 at position 42.2

Molecular Location: base pairs 231,241,173 to 231,277,973 on chromosome 1 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- acyl-CoA:dihydroxyacetonephosphateacyltransferase
- DAP-AT
- DAPAT
- DHAP-AT
- DHAPAT
- dihydroxyacetone phosphate acyltransferase
- glycerone-phosphate O-acyltransferase
- GNPAT_HUMAN

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: The Biogenesis and Cell Biology of Peroxisomes in Human Health and Disease https://www.ncbi.nlm.nih.gov/books/NBK6339/
- The Cell: A Molecular Approach (second edition, 2000): Peroxisomes https://www.ncbi.nlm.nih.gov/books/NBK9930/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GNPAT%5BTIAB%5D%29+OR+%28glyceronephosphate+O-acyltransferase%5BTIAB%5D%29%29+OR+%28%28dihydroxyacetone+phosphate+acyltransferase%5BTIAB%5D%29+OR+%28DHAPAT%5BTIAB%5D%29+OR+%28dihydroxyacetonephosphate+acyltransferase%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+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- GLYCERONEPHOSPHATE O-ACYLTRANSFERASE
  http://omim.org/entry/602744

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GNPAT%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:4416
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:8443
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8443
- UniProt
  https://www.uniprot.org/uniprot/O15228

Sources for This Summary

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