

## 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.

### Other Names for This Gene

- acyl-CoA:dihydroxyacetonephosphate acyltransferase
- DAP-AT
- DAPAT

- DHAP-AT
- DHAPAT
- dihydroxyacetone phosphate acyltransferase
- glycerone-phosphate O-acyltransferase
- GNPAT HUMAN

## Additional Information & Resources

## Tests Listed in the Genetic Testing Registry

- Tests of GNPAT ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8443\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8443[geneid]))

## Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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; GNPAT (<https://omim.org/entry/602744>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8443>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GNPAT\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GNPAT[gene]))

## References

- Liu D, Nagan N, Just WW, Rodemer C, Thai TP, Zoeller RA. Role of dihydroxyacetonephosphate acyltransferase in the biosynthesis of plasmalogens and nonether glycerolipids. *J Lipid Res.* 2005 Apr;46(4):727-35. doi:10.1194/jlr.M400364-JLR200. Epub 2005 Feb 1. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15687349>)
- Ofman R, Hettema EH, Hogenhout EM, Caruso U, Muijsers AO, Wanders RJ. Acyl-CoA: dihydroxyacetonephosphate acyltransferase: cloning of the human cDNA and resolution of the molecular basis in rhizomelic chondrodysplasia punctata type 2.

Hum Mol Genet. 1998 May;7(5):847-53. doi: 10.1093/hmg/7.5.847. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/9536089>)

- Ofman R, Lajmir S, Wanders RJ. Etherphospholipid biosynthesis and dihydroxyacetone-phosphate acyltransferase: resolution of the genomic organization of the human *gnpat* gene and its use in the identification of novel mutations. Biochem Biophys Res Commun. 2001 Mar 2;281(3):754-60. doi:10.1006/bbrc.2001.4407. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11237722>)
- Thai TP, Rodemer C, Jauch A, Hunziker A, Moser A, Gorgas K, Just WW. Impaired membrane traffic in defective ether lipid biosynthesis. Hum Mol Genet. 2001 Jan 15;10(2):127-36. doi: 10.1093/hmg/10.2.127. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11152660>)
- Wanders RJ, Schumacher H, Heikoop J, Schutgens RB, Tager JM. Human dihydroxyacetonephosphate acyltransferase deficiency: a new peroxisomal disorder. J Inher Metab Dis. 1992;15(3):389-91. doi: 10.1007/BF02435984. No abstract available. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/1405476>)

## Genomic Location

The *GNPAT* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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