AGPS gene

alkylglycerone phosphate synthase

Normal Function

The *AGPS* gene provides instructions for making an enzyme known as alkylglycerone phosphate synthase. 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, alkylglycerone phosphate synthase is responsible for a critical 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 three mutations in the *AGPS* gene have been found to cause rhizomelic chondrodysplasia punctata type 3 (RCDP3). These mutations change single protein building blocks (amino acids) in alkylglycerone phosphate synthase, which alters the structure of the enzyme and significantly reduces its activity. A shortage of functional alkylglycerone phosphate synthase 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 RCDP3.

Other Names for This Gene

- ADAP-S
- ADAS_HUMAN
- ADHAPS

- alkyl-DHAP synthase
- alkyl-dihydroxyacetone phosphate synthase
- alkyldihydroxyacetone phosphate synthetase
- alkyldihydroxyacetonephosphate synthase, peroxisomal
- alkylglycerone-phosphate synthase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AGPS (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8540[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28AGPS%5BTIAB%5D%29+OR+%28alkylglycerone+phosphate+synthase%5BTIAB%5D%29%29+OR+%28%28alkyl-dihydroxyacetonephosphate+synthase%5BTIAB%5D%29+OR+%28alkyldihydroxyacetonephosphate+synthase%5BTIAB%5D%29+OR+%28alkyl-dihydroxyacetone+phosphate+synthase%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)

Catalog of Genes and Diseases from OMIM

- ALKYLGLYCERONE-PHOSPHATE SYNTHASE; AGPS (https://omim.org/entry/603051)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/8540)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AGPS[gene])

References

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Genomic Location

The *AGPS* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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