

ALPL gene

alkaline phosphatase, biomineralization associated

Normal Function

The *ALPL* gene provides instructions for making an enzyme called tissue-nonspecific alkaline phosphatase (TNSALP). This enzyme plays an important role in the growth and development of bones and teeth. It is also active in many other tissues, particularly in the liver and kidneys. This enzyme acts as a phosphatase, which means that it removes clusters of oxygen and phosphorus atoms (phosphate groups) from other molecules.

TNSALP is essential for the process of mineralization, in which minerals such as calcium and phosphorus are deposited in developing bones and teeth. Mineralization is critical for the formation of bones that are strong and rigid and teeth that can withstand chewing and grinding.

Health Conditions Related to Genetic Changes

Hypophosphatasia

About 300 mutations in the *ALPL* gene have been identified in people with hypophosphatasia. Most of these mutations change a single protein building block (amino acid) in TNSALP. Other mutations insert or delete genetic material in the *ALPL* gene or change the way the gene's instructions are used to build the enzyme.

Mutations in the *ALPL* gene lead to the production of an abnormal version of TNSALP that cannot participate effectively in the mineralization of developing bones and teeth. A shortage of functional TNSALP allows substances that are normally processed by the enzyme to build up in the body. Researchers believe that a buildup of one of these compounds, inorganic pyrophosphate, underlies the defective mineralization of bones and teeth in people with hypophosphatasia.

ALPL mutations that almost completely eliminate the activity of TNSALP usually result in the more severe forms of hypophosphatasia. Other mutations, which reduce but do not eliminate the activity of the enzyme, are often responsible for milder forms of the condition.

Other Names for This Gene

- alkaline phosphatase, liver/bone/kidney
- alkaline phosphomonoesterase
- AP-TNAP
- glycerophosphatase
- HOPS
- MGC161443
- PPBT_HUMAN
- tissue non-specific alkaline phosphatase
- tissue-nonspecific ALP
- TNALP
- TNAP
- TNSALP

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ALPL ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=249\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=249[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ALPL%5BTIAB%5D%29+OR+%28alkaline+phosphatase+AND+hypophosphatasia%5BTIAB%5D%29%29+OR+%28%28tissue+nonspecific+alkaline+phosphatase%5BTIAB%5D%29+OR+%28tissue+non-specific+alkaline+phosphatase%5BTIAB%5D%29+OR+%28TNAP%5BTIAB%5D%29+OR+%28TNSALP%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+1800+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- ALKALINE PHOSPHATASE, LIVER; ALPL (<https://omim.org/entry/171760>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/249>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ALPL\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ALPL[gene]))

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Genomic Location

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

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