

PNPO gene

pyridoxamine 5'-phosphate oxidase

Normal Function

The *PNPO* gene provides instructions for producing an enzyme called pyridox(am)ine 5'-phosphate oxidase. This enzyme is involved in the breakdown (metabolism) of vitamin B6. Specifically, it chemically modifies two forms of vitamin B6 obtained from food (pyridoxine and pyridoxamine) to create pyridoxal 5'-phosphate (PLP). PLP is the active form of vitamin B6, and it is necessary for many chemical reactions in the body, including protein metabolism and the processing of chemicals that transmit signals in the brain (neurotransmitters). Pyridox(am)ine 5'-phosphate oxidase is active (expressed) in cells throughout the body, with the highest amounts found in the liver.

Health Conditions Related to Genetic Changes

Pyridoxal phosphate-responsive seizures

Several variants (also called mutations) in the *PNPO* gene have been found to cause pyridoxal phosphate-responsive seizures (sometimes called pyridoxamine 5'-phosphate oxidase deficiency or PNPO deficiency). People with PNPO deficiency typically develop seizures shortly after birth. Most of the variants in the *PNPO* gene that cause PNPO deficiency change one protein building block (amino acid) in the pyridox(am)ine 5'-phosphate oxidase enzyme, impairing its normal function. The resulting enzyme cannot effectively metabolize vitamin B6 to produce PLP. A shortage of PLP can disrupt the function of many other proteins and enzymes that need PLP in order to be effective. Because PLP plays an important role in neurotransmitter metabolism, a lack of PLP is believed to cause the seizures that are characteristic of PNPO deficiency.

Other Names for This Gene

- HEL-S-302
- PDXPO
- pyridoxal 5'-phosphate oxidase
- pyridoxal 5'-phosphate synthetase
- pyridoxamine-phosphate oxidase
- pyridoxine 5'-phosphate oxidase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PNPO ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55163\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55163[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PNPO%29+OR+%28pyridoxamine+5%27;-phosphate+oxidase%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%29%29>)

Catalog of Genes and Diseases from OMIM

- PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE; PNPO (<https://omim.org/entry/603287>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/55163>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PNPO\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PNPO[gene]))

References

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Genomic Location

The *PNPO* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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