# TYMP gene

thymidine phosphorylase

## Normal Function

The *TYMP* gene (previously known as *ECGF1*) provides instructions for making an enzyme called thymidine phosphorylase. Thymidine is a molecule known as a nucleoside. After a chemical modification, thymidine is used as a building block of DNA. Thymidine phosphorylase converts thymidine into two smaller molecules, 2-deoxyribose 1-phosphate and thymine. By breaking down thymidine, this chemical reaction helps regulate the level of nucleosides in cells.

Thymidine phosphorylase plays an important role in maintaining the appropriate amount of thymidine in cell structures called mitochondria. Mitochondria convert the energy from food into a form that cells can use. Although most DNA is packaged in chromosomes within the nucleus, mitochondria also have a small amount of their own DNA, called mitochondrial DNA or mtDNA. Mitochondria use nucleosides, including thymidine, to build new molecules of mtDNA.

## Health Conditions Related to Genetic Changes

Mitochondrial neurogastrointestinal encephalopathy disease

About 50 variants (also called mutations) in the *TYMP* gene have been identified in people with mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease. This condition affects several parts of the body, particularly the digestive system and nervous system.

*TYMP* gene variants greatly reduce or eliminate the activity of thymidine phosphorylase. A shortage of this enzyme allows thymidine to build up to very high levels in the body. This extra thymidine can damage mtDNA, disrupting its usual maintenance and repair processes. As a result, variants can accumulate in mtDNA, causing it to become unstable. In people with MNGIE disease, mitochondria may also have less mtDNA than usual (mtDNA depletion). These genetic changes impair the normal function of mitochondria. Although mtDNA abnormalities underlie the digestive and neurological problems that are characteristic of MNGIE disease, it is unclear how defective mitochondria cause the specific features of the disorder.

## Other Names for This Gene

- ECGF1
- endothelial cell growth factor 1 (platelet-derived)
- gliostatin
- hPD-ECGF
- MNGIE
- PD-ECGF
- PDECGF
- TdRPase
- TP
- TYPH_HUMAN

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TYMP (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1890[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TYMP+NOT+tympanometry+NOT+otitis%5BTIAB%5D%29+OR+%28thymidine+phosphorylase%5BTIAB%5D%29+OR+%28ECGF1%5BTIAB%5D%29+OR+%28MNGIE%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

- THYMIDINE PHOSPHORYLASE; TYMP (https://omim.org/entry/131222)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1890)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TYMP[gene])

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## Genomic Location

The *TYMP* gene is found on chromosome 22 (https://medlineplus.gov/genetics/chromos

ome/22/).

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