

CLPP gene

caseinolytic mitochondrial matrix peptidase proteolytic subunit

Normal Function

The *CLPP* gene provides instructions for making the ClpP subunit protein. Multiple copies of this protein interact with each other to form a barrel-shaped chamber known as the ClpP complex. This complex is one of two parts of the ClpXP protease, which breaks down abnormally folded proteins. The other part of the ClpXP protease, called the ClpX complex, unfolds the abnormal proteins and feeds them into the chamber formed by the ClpP complex, where they are broken down into small fragments.

The ClpP complex is found in structures in the cell called mitochondria, which are the energy-producing centers of cells.

Health Conditions Related to Genetic Changes

Perrault syndrome

At least three mutations in the *CLPP* gene have been found in families with Perrault syndrome, a condition characterized by hearing loss in affected males and females and abnormalities of the ovaries in affected females. The *CLPP* gene mutations involved in Perrault syndrome likely alter the structure of the barrel-shaped chamber formed by the ClpP complex. These changes may impair the breakdown of misfolded mitochondrial proteins, which could impact mitochondrial function; however, it is unclear how these changes lead to the features of Perrault syndrome.

Other Names for This Gene

- ATP-dependent Clp protease proteolytic subunit, mitochondrial
- ATP-dependent protease ClpAP, proteolytic subunit, human
- ClpP caseinolytic peptidase ATP-dependent, proteolytic subunit
- ClpP caseinolytic protease, ATP-dependent, proteolytic subunit homolog
- endopeptidase Clp
- PRLTS3
- putative ATP-dependent Clp protease proteolytic subunit, mitochondrial

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CLPP ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8192\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8192[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28CLPP%5BTIAB%5D%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

- CASEINOLYTIC MITOCHONDRIAL MATRIX PEPTIDASE PROTEOLYTIC SUBUNIT; CLPP (<https://omim.org/entry/601119>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8192>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CLPP\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CLPP[gene]))

References

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Genomic Location

The *CLPP* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

Last updated December 1, 2014