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

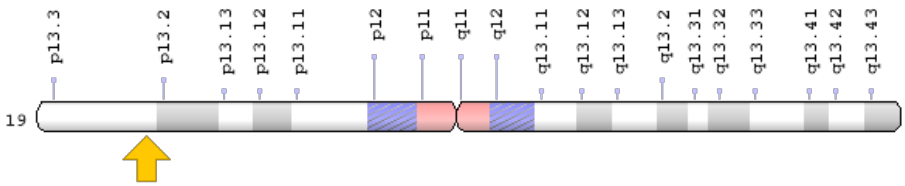
Perrault syndrome

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.

Chromosomal Location

Cytogenetic Location: 19p13.3, which is the short (p) arm of chromosome 19 at position 13.3

Molecular Location: base pairs 6,361,531 to 6,370,242 on chromosome 19 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Clinical Information from GeneReviews

- Perrault Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK242617

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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
  http://omim.org/entry/601119

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CLPP.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CLPP%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2084
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:8192

- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8192
- UniProt
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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/CLPP

Reviewed: December 2014
Published: November 12, 2019

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services