CTSA gene

cathepsin A

Normal Function

The *CTSA* gene provides instructions for making a protein called cathepsin A. Cathepsin A can act as a protease, cutting apart other proteins to break them down. Cathepsin A can also act as a protective protein, interacting with other enzymes to prevent them from breaking down prematurely. Based on this protective function, this enzyme is also called protective protein/cathepsin A or PPCA.

Cathepsin A is active in cellular compartments called lysosomes. These compartments contain enzymes that digest and recycle materials when they are no longer needed. Cathepsin A works together with the enzymes beta-galactosidase and neuraminidase 1, which play a role in the breakdown of sugar molecules (oligosaccharides) attached to certain proteins (glycoproteins) or fats (glycolipids).

On the cell surface, cathepsin A forms a complex with neuraminidase 1 and elastin-binding protein, creating the elastin-binding protein receptor. This receptor complex plays a role in the formation of elastic fibers, which are components of the connective tissues that make up the body's supportive framework.

Health Conditions Related to Genetic Changes

Galactosialidosis

Variants (also called mutations) in the *CTSA* gene have been found to cause galactosialidosis, a condition that affects many areas of the body. There are three forms of galactosialidosis that are distinguished by the age at which symptoms develop and the pattern of features. The features of the condition vary, but often include problems with the heart, skeleton, vision, hearing, and other systems.

Most of the CTSA gene variants that cause galactosialidosis change one protein building block (amino acid) in cathepsin A. In the Japanese population, the most common variant (written as SpDEx7) disrupts how the gene's instructions are used to make the protein.

Many *CTSA* gene variants disrupt the protein structure of cathepsin A, impairing its ability to join with neuraminidase 1 and beta-galactosidase or elastin-binding protein. As a result, these other enzymes are not functional, or they break down prematurely. It is

not well understood how the loss of these four proteins causes the signs and symptoms of galactosialidosis.

Other Names for This Gene

- beta-galactosidase 2
- beta-galactosidase protective protein
- GSL
- PPCA
- PPGB
- PPGB_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CTSA (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5476[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CTSA%5BTIAB%5D%29+OR+%28cathepsin+A%5BTIAB%5D%29%29+OR+%28PPCA%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+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CATHEPSIN A; CTSA (https://omim.org/entry/613111)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5476)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CTSA[gene])

References

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Genomic Location

The *CTSA* gene is found on chromosome 20 (https://medlineplus.gov/genetics/chromosome/20/).

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