

## PRSS1 gene

serine protease 1

### Normal Function

The *PRSS1* gene provides instructions for making an enzyme called cationic trypsinogen. This enzyme is a serine peptidase, which is a type of enzyme that cuts (cleaves) other proteins into smaller pieces. Cationic trypsinogen is produced in the pancreas and helps with the digestion of food. Cationic trypsinogen is secreted by the pancreas and transported to the small intestine, where it is cleaved to form trypsinogen. When the enzyme is needed, trypsinogen is cleaved again into its working (active) form called trypsin. Trypsin aids in digestion by cutting protein chains at the protein building blocks (amino acids) arginine or lysine, which breaks down the protein. Trypsin also turns on (activates) other digestive enzymes that are produced in the pancreas to further facilitate digestion.

A particular region of trypsin is attached (bound) to a calcium molecule. As long as trypsin is bound to calcium, the enzyme is protected from being broken down. When digestion is complete and trypsin is no longer needed, the calcium molecule is removed from the enzyme, which allows trypsin to be broken down.

### Health Conditions Related to Genetic Changes

#### Hereditary pancreatitis

More than 40 mutations in the *PRSS1* gene have been found to cause hereditary pancreatitis, a condition characterized by recurrent episodes of inflammation of the pancreas (pancreatitis), which can lead to a loss of pancreatic function. Most of these mutations change single protein building blocks (amino acids) in cationic trypsinogen. Some *PRSS1* gene mutations result in the production of a cationic trypsinogen enzyme that is prematurely converted to trypsin while it is still in the pancreas. Other mutations prevent trypsin from being broken down. The most common *PRSS1* gene mutation that causes hereditary pancreatitis replaces the amino acid arginine with the amino acid histidine at position 122 in the enzyme (written Arg122His or R122H). As a result of this mutation, the enzyme is not able to be broken down, even when it is no longer bound to calcium.

Trypsin activity in the pancreas can damage pancreatic tissue and can also trigger an immune response, causing inflammation in the pancreas and leading to episodes of

pancreatitis.

### Other Names for This Gene

- beta-trypsin
- cationic trypsinogen
- protease, serine 1
- protease, serine, 1 (trypsin 1)
- TRP1
- TRY1
- TRY1\_HUMAN
- TRY4
- TRYP1
- trypsin-1
- trypsin-1 preproprotein
- trypsinogen 1
- trypsinogen A

### Additional Information & Resources

#### Tests Listed in the Genetic Testing Registry

- Tests of PRSS1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5644\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5644[geneid]))

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PRSS1%5BTIAB%5D%29+OR+%28cationic+trypsinogen%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%29%29%29>)

#### Catalog of Genes and Diseases from OMIM

- PROTEASE, SERINE, 1; PRSS1 (<https://omim.org/entry/276000>)

#### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5644>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PRSS1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PRSS1[gene]))

## References

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- Rebours V, Levy P, Ruzsiewicz P. An overview of hereditary pancreatitis. *Dig Liver Dis*. 2012 Jan;44(1):8-15. doi: 10.1016/j.dld.2011.08.003. Epub 2011 Sep 9. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21907651>)

## Genomic Location

The *PRSS1* gene is found on chromosome 7 (<https://medlineplus.gov/genetics/chromosome/7/>).

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