

## PEPD gene

peptidase D

### Normal Function

The *PEPD* gene provides instructions for making the enzyme prolidase, also called peptidase D. Prolidase helps divide certain dipeptides, which are molecules composed of two protein building blocks (amino acids). Specifically, prolidase divides dipeptides containing the amino acids proline or hydroxyproline. By freeing these amino acids, prolidase helps make them available for use in producing proteins that the body needs.

Prolidase is also involved in the final step of the breakdown of some proteins obtained through the diet and proteins that are no longer needed in the body. Prolidase is particularly important in the breakdown of collagens, a family of proteins that are rich in proline and hydroxyproline. Collagens are an important part of the extracellular matrix, which is the lattice of proteins and other molecules outside the cell. The extracellular matrix strengthens and supports connective tissues, such as skin, bone, cartilage, tendons, and ligaments. Collagen breakdown occurs during the maintenance (remodeling) of the extracellular matrix.

### Health Conditions Related to Genetic Changes

#### Prolidase deficiency

At least 19 mutations in the *PEPD* gene have been identified in people with prolidase deficiency, a disorder with a wide variety of signs and symptoms including skin problems and intellectual disability. The *PEPD* gene mutations identified in people with prolidase deficiency result in the loss of prolidase enzyme activity.

It is not well understood how the absence of prolidase activity results in the various signs and symptoms of prolidase deficiency. Researchers have suggested that accumulation of dipeptides that have not been broken down may lead to cell death. When cells die, their contents are released into the surrounding tissue, which could cause inflammation and lead to the skin problems seen in prolidase deficiency. Impaired collagen breakdown during remodeling of the extracellular matrix may also contribute to the skin problems. The intellectual disability that occurs in prolidase deficiency might result from problems in processing neuropeptides, which are brain signaling proteins that are rich in proline. It is unclear how absence of prolidase activity results in the other features of prolidase deficiency.

## Other Names for This Gene

- aminoacyl-L-proline hydrolase
- imidodipeptidase
- MGC10905
- PEPD\_HUMAN
- PROLIDASE
- proline dipeptidase
- X-Pro dipeptidase
- xaa-Pro dipeptidase

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of PEPD ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5184\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5184[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PEPD%5BTIAB%5D%29+OR+%28peptidase+D%5BTIAB%5D%29%29+OR+%28%28PROLIDASE%5BTIAB%5D%29+OR+%28xaa-Pro+dipeptidase%5BTIAB%5D%29+OR+%28imidodipeptidase%5BTIAB%5D%29+OR+%28X-Pro+dipeptidase%5BTIAB%5D%29+OR+%28proline+dipeptidase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28paroxysmal+extreme+pain+disorder%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- PEPTIDASE D; PEPD (<https://omim.org/entry/613230>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5184>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PEPD\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PEPD[gene]))

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## Genomic Location

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

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