

## PROP1 gene

PROP paired-like homeobox 1

### Normal Function

The *PROP1* gene provides instructions for making a protein that helps control the activity of many other genes. On the basis of this action, the PROP1 protein is known as a transcription factor. This protein is found only in the pituitary gland, which is located at the base of the brain. The pituitary gland releases hormones needed for growth, reproduction, and other critical body functions. The PROP1 protein helps in the specialization (differentiation) of cell types within the pituitary gland.

### Health Conditions Related to Genetic Changes

#### Combined pituitary hormone deficiency

At least 25 mutations in the *PROP1* gene have been found to cause combined pituitary hormone deficiency. The most common mutation deletes two DNA building blocks in the *PROP1* gene (written as 301-302delAG). This deletion leads to the production of an abnormally short protein that cannot function properly. The shortened protein is less able to control the activity of other genes, which reduces pituitary cell differentiation and prevents the release of hormones from the pituitary gland. A lack of these hormones can cause short stature, delayed or absent puberty, and other signs and symptoms of combined pituitary hormone deficiency.

### Other Names for This Gene

- homeobox protein prophet of Pit-1
- pituitary-specific homeodomain factor
- PROP-1
- PROP1\_HUMAN
- prophet of Pit1, paired-like homeodomain transcription factor

### Additional Information & Resources

#### Tests Listed in the Genetic Testing Registry

- Tests of PROP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5626\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5626[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28PROP1%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

- PROP PAIRED-LIKE HOMEBOX 1; PROP1 (<https://omim.org/entry/601538>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5626>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PROP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PROP1[gene]))

## **References**

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- Kelberman D, Turton JP, Woods KS, Mehta A, Al-Khawari M, Greening J, Swift PG, Otonkoski T, Rhodes SJ, Dattani MT. Molecular analysis of novel PROP1 mutations associated with combined pituitary hormone deficiency (CPHD). *Clin Endocrinol (Oxf).* 2009 Jan;70(1):96-103. doi: 10.1111/j.1365-2265.2008.03326.x. Epub 2008 Jun 25. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19128366>)
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## **Genomic Location**

The *PROP1* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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