Genetics Home Reference

Your Guide to Understanding Genetic Conditions

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

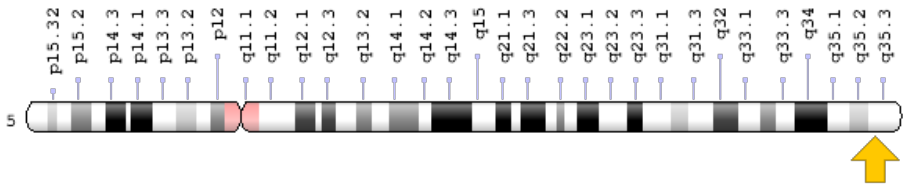
combined pituitary hormone deficiency

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.

Chromosomal Location

Cytogenetic Location: 5q35.3, which is the long (q) arm of chromosome 5 at position 35.3

Molecular Location: base pairs 177,992,235 to 177,996,242 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Educational Resources

- Endocrinology: An Integrated Approach (first edition, 2001): The Pituitary Gland
  https://www.ncbi.nlm.nih.gov/books/NBK27/

GeneReviews

- PROP1-Related Combined Pituitary Hormone Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1347

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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

OMIM

- PROP PAIRED-LIKE HOMEOBOX 1
  http://omim.org/entry/601538

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PROP1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PROP1%5Bgene%5D
- HGNC Gene Family: PRD class homeoboxes and pseudogenes
  http://www.genenames.org/cgi-bin/genefamilies/set/521
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9455

- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5626
- UniProt
  http://www.uniprot.org/uniprot/O75360

Sources for This Summary

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