ENG gene

endoglin

Normal Function

The *ENG* gene provides instructions for making a protein called endoglin. This protein is found on the surface of cells, especially in the lining of developing arteries. It forms a complex with growth factors and other proteins involved in the development of blood vessels. In particular, this complex is involved in the specialization of new blood vessels into arteries or veins.

Health Conditions Related to Genetic Changes

Hereditary hemorrhagic telangiectasia

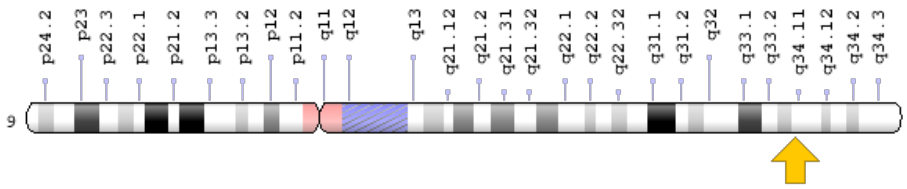
Dozens of mutations in the *ENG* gene have been found to cause hereditary hemorrhagic telangiectasia type 1. Many *ENG* gene mutations substitute one protein building block (amino acid) for another amino acid in the endoglin protein, which impairs the protein's function. Other mutations prevent production of the endoglin protein or result in an abnormally small protein that cannot function. The shortage of functional endoglin appears to interfere with the development of boundaries between arteries and veins, resulting in the signs and symptoms of hereditary hemorrhagic telangiectasia type 1.

Pulmonary arterial hypertension

Chromosomal Location

Cytogenetic Location: 9q34.11, which is the long (q) arm of chromosome 9 at position 34.11

Molecular Location: base pairs 127,815,012 to 127,854,773 on chromosome 9 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CD105
- EGLN_HUMAN
- END
- endoglin (Osler-Rendu-Weber syndrome 1)
- endoglin precursor
- HHT1
- ORW
- ORW1
- Transforming Growth Factor P Receptor III

Additional Information & Resources

Clinical Information from GeneReviews

- Hereditary Hemorrhagic Telangiectasia
  https://www.ncbi.nlm.nih.gov/books/NBK1351

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ENG%5BTIAB%5D%29+OR+%28endoglin%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

Catalog of Genes and Diseases from OMIM

- ENDOGLIN
  http://omim.org/entry/131195

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/ENGID40452ch9q34.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ENG%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3349
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2022

- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2022
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Reviewed: February 2007
Published: September 10, 2019

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