ACVRL1 gene

activin A receptor like type 1

Normal Function

The *ACVRL1* gene provides instructions for making a protein called activin receptor-like kinase 1. This protein is found on the surface of cells, especially in the lining of developing arteries.

The ACVRL1 protein is a receptor. It acts as a "lock" waiting for a specific protein, called its ligand, to serve as the "key." In the case of the ACVRL1 protein, the ligand is called transforming growth factor beta. The interaction between these proteins plays a role in the development of blood vessels. In particular, this protein interaction 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 *ACVRL1* gene have been found to cause hereditary hemorrhagic telangiectasia type 2. Many *ACVRL1* gene mutations substitute one protein building block (amino acid) for another amino acid in the ACVRL1 protein, which impairs the protein's function. Other mutations prevent production of the ACVRL1 protein or result in an abnormally small protein that cannot function. The shortage of functional ACVRL1 protein appears to interfere with the development of boundaries between arteries and veins, resulting in the signs and symptoms of hereditary hemorrhagic telangiectasia type 2.

Pulmonary arterial hypertension

MedlinePlus Genetics provides information about Pulmonary arterial hypertension

Other disorders

A common genetic variation (polymorphism) in the *ACVRL1* gene has been found to appear more often in people who develop arteriovenous malformations in the brain, but who do not have other signs or symptoms of hereditary hemorrhagic telangiectasia, than in the general population. The polymorphism replaces a particular DNA building block (nucleotide) called adenine with the nucleotide guanine (written as IVS3-35 A>

G). This genetic variation may affect the development of blood vessels in the brain, resulting in an increased risk of arteriovenous malformations.

Other Names for This Gene

- activin A receptor type II-like 1
- activin A receptor type IL
- Activin A receptor, type II-like kinase 1
- ACVL1_HUMAN
- ACVRLK1
- ALK-1
- ALK1
- EC 2.7.1.37
- HHT
- HHT2
- ORW2
- Serine/threonine-protein kinase Receptor R3 Precursor
- SKR3
- TGF-B Superfamily Receptor Type I

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ACVRL1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=94[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ACVRL1%5BTIAB%5D%29+OR+%28activin+A+receptor+type+II-like+1%5BTIAB%5D%29%29+OR+%28%28HHT%5BTIAB%5D%29+OR+%28ALK1%5BTIAB%5D%29+OR+%28HHT2%5BTIAB%5D%29+OR+%28ORW2%5BTIAB%5D%29+OR+%28SKR3%5BTIAB%5D%29+OR+%28ALK-1%5BTIAB%5D%29+OR+%28ACVRLK1%5BTIAB%5D%29+OR+%28Activin+A+receptor,+type+II-like+kinase+1%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ARTERIOVENOUS MALFORMATIONS OF THE BRAIN (https://omim.org/entry/10

8010)

- ACTIVIN A RECEPTOR, TYPE II-LIKE 1; ACVRL1 (https://omim.org/entry/601284)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/94)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ACVRL1[gene])

References

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Genomic Location

The *ACVRL1* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chrom

osome/12/).

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