



BMPR1A gene

bone morphogenetic protein receptor type 1A

Normal Function

The *BMPR1A* gene provides instructions for making a protein called bone morphogenetic protein receptor 1A. This receptor protein has a specific site into which certain other proteins, called ligands, fit like keys into locks. Specifically, the BMPR1A protein attaches (binds) to ligands in the transforming growth factor beta (TGF- β) pathway. This signaling pathway allows the environment outside the cell to affect how the cell produces other proteins. The BMPR1A receptor protein and its ligands are involved in transmitting chemical signals from the cell membrane to the nucleus.

When the BMPR1A protein is bound to a ligand, it turns on (activates) a group of related proteins (a protein complex) called SMAD proteins. The activated SMAD protein complex is then transported into the cell's nucleus, where it regulates cell growth and division (proliferation) and the activity of particular genes.

Health Conditions Related to Genetic Changes

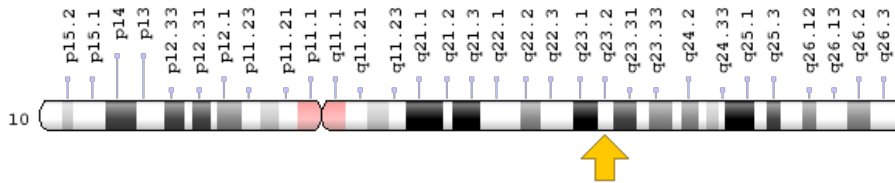
Juvenile polyposis syndrome

More than 60 mutations in the *BMPR1A* gene have been found to cause juvenile polyposis syndrome. Most *BMPR1A* gene mutations result in the production of an abnormally short, nonfunctional protein. As a result, the BMPR1A protein cannot bind to ligands in the TGF- β pathway. This disruption in binding interferes with the activation of the SMAD protein complex. This inactive complex is not transported to the nucleus, where it is needed to regulate cell growth and the activity of certain genes. Unregulated cell growth can lead to polyp formation in people with juvenile polyposis syndrome.

Chromosomal Location

Cytogenetic Location: 10q23.2, which is the long (q) arm of chromosome 10 at position 23.2

Molecular Location: base pairs 86,755,786 to 86,927,969 on chromosome 10 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- activin A receptor, type II-like kinase 3
- ACVRLK3
- ALK3
- BMR1A_HUMAN
- bone morphogenetic protein receptor type IA
- bone morphogenetic protein receptor, type IA
- bone morphogenetic protein receptor, type IA precursor
- CD292
- serine/threonine-protein kinase receptor R5
- SKR5

Additional Information & Resources

Educational Resources

- Cancer Medicine (sixth edition, 2003): Familial Juvenile Polyposis Coli
<https://www.ncbi.nlm.nih.gov/books/NBK12959/#A4571>
- Developmental Biology (sixth edition, 2000): The Smad pathway activated by TGF- β superfamily ligands
<https://www.ncbi.nlm.nih.gov/books/NBK10043/figure/A1057/>

Clinical Information from GeneReviews

- Juvenile Polyposis Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1469>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28BMPR1A%5BTIAB%5D%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE IA
<http://omim.org/entry/601299>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_BMPR1A.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=BMPR1A%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:1076
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:657>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/657>
- UniProt
<https://www.uniprot.org/uniprot/P36894>

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<https://ghr.nlm.nih.gov/gene/BMPR1A>

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