BMPR2 gene

bone morphogenetic protein receptor type 2

Normal Function

The *BMPR2* gene provides instructions for making a protein called bone morphogenetic protein receptor type 2. The *BMPR2* gene belongs to a family of genes originally identified for its role in regulating the growth and maturation (differentiation) of bone and cartilage. Recently, researchers have found that this gene family plays a broader role in regulating the growth and differentiation of numerous types of cells.

Bone morphogenetic protein receptor type 2 spans the cell membrane, so that one end of the protein is on the outer surface of the cell and the other end remains inside the cell. This positioning allows the protein to receive and transmit signals that help the cell respond to its environment by growing and dividing (cell proliferation) or by undergoing controlled cell death (apoptosis). This balance of cell proliferation and apoptosis regulates the number of cells in tissues.

Health Conditions Related to Genetic Changes

pulmonary arterial hypertension

Researchers have identified more than 350 *BMPR2* gene mutations that can cause pulmonary arterial hypertension, a condition characterized by abnormally high blood pressure (hypertension) in the blood vessel that carries blood from the heart to the lungs (the pulmonary artery). About half of the mutations involved in this condition disrupt the assembly of bone morphogenetic protein receptor type 2, reducing the amount of this protein in cells. Other mutations prevent bone morphogenetic protein receptor type 2 from reaching the cell surface or alter its structure so it cannot receive or transmit signals.

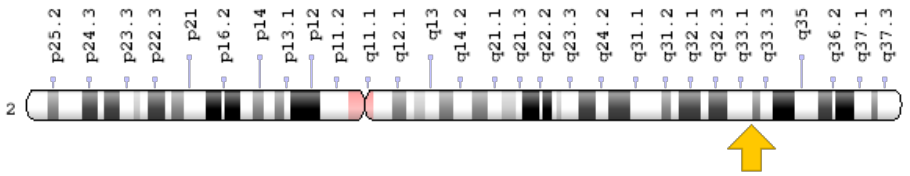
It remains unclear how *BMPR2* gene mutations cause pulmonary arterial hypertension. Researchers suggest that a mutation in this gene promotes cell proliferation or prevents cell death, resulting in an overgrowth of cells in the smallest arteries throughout the lungs. As a result, these arteries narrow in diameter, which increases the resistance to blood flow through the lungs. To overcome the increased resistance, blood pressure increases in the pulmonary artery and in the right ventricle of the heart, which is the chamber that pumps blood into the pulmonary artery. Signs and symptoms of pulmonary arterial hypertension occur when increased blood pressure cannot fully overcome the elevated resistance, and blood flow to the body is insufficient.

pulmonary veno-occlusive disease

Chromosomal Location

Cytogenetic Location: 2q33.1-q33.2, which is the long (q) arm of chromosome 2 between positions 33.1 and 33.2

Molecular Location: base pairs 202,376,310 to 202,567,751 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BMPR-II
- BMPR2_HUMAN
- BMPR3
- BMR2
- bone morphogenetic protein receptor type II
- bone morphogenetic protein receptor, type II (serine/threonine kinase)
- BRK-3
- PPH1
- Receptor, Type II BMP
- serine/threonine kinase
- T-ALK
- type II activin receptor-like kinase

Additional Information & Resources

GeneReviews

- Heritable Pulmonary Arterial Hypertension
  https://www.ncbi.nlm.nih.gov/books/NBK1485

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28BMPR2%5BTIAB%5D%29+OR+%28BMPR-II%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE II
  http://omim.org/entry/600799

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_BMPR2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=BMPR2%5Bgene%5D
- HGNC Gene Family: Type 2 receptor serine/threonine kinases
  https://www.genenames.org/cgi-bin/genefamilies/set/346
- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1078
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/659
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