

RBPJ gene

recombination signal binding protein for immunoglobulin kappa J region

Normal Function

The *RBPJ* gene provides instructions for making a protein called RBP-J, which is an integral part of a signaling pathway known as the Notch pathway. Notch signaling controls how certain types of cells develop in the growing embryo, including those that form the bones, heart, muscles, nerves, and blood. Signaling through the Notch pathway stimulates the RBP-J protein to attach (bind) to specific regions of DNA and control the activity of genes that play a role in cellular development.

Health Conditions Related to Genetic Changes

Adams-Oliver syndrome

Mutations in the *RBPJ* gene lead to Adams-Oliver syndrome, a condition characterized by areas of missing skin (aplasia cutis congenita), usually on the scalp, and malformations of the hands and feet. These mutations change single protein building blocks in the RBP-J protein, altering the region of the protein that normally binds to DNA.

The altered proteins are unable to bind to DNA, preventing the activation of particular genes. These changes in gene activity impair the proper development of the skin on the top of the head and the bones in the hands and feet. It is unclear why impaired development primarily affects these tissues.

Rheumatoid arthritis

MedlinePlus Genetics provides information about Rheumatoid arthritis

Other Names for This Gene

- AOS3
- CBF-1
- CBF1
- csl
- H-2K binding factor-2
- IGKJRB

- IGKJRB1
- immunoglobulin kappa J region recombination signal binding protein 1
- KBF2
- RBP-J
- RBP-J kappa
- RBP-JK
- RBPJK
- RBPSUH
- recombining binding protein suppressor of hairless
- recombining binding protein suppressor of hairless isoform 3
- renal carcinoma antigen NY-REN-30
- SUH
- SUH_HUMAN
- suppressor of hairless homolog

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RBPJ ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3516\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3516[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RBPJ%5BTIAB%5D%29+OR+%28recombination+signal+binding+protein+for+immunoglobulin+kappa+J+region%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bm%5D+AND+%22last+1800+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- RECOMBINATION SIGNAL-BINDING PROTEIN FOR IMMUNOGLOBULIN KAPPA J REGION; RBPJ (<https://omim.org/entry/147183>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3516>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RBPJ\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RBPJ[gene]))

References

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- Hassed SJ, Wiley GB, Wang S, Lee JY, Li S, Xu W, Zhao ZJ, Mulvihill JJ, Robertson J, Warner J, Gaffney PM. RBPJ mutations identified in two families affected by Adams-Oliver syndrome. *Am J Hum Genet.* 2012 Aug 10;91(2):391-5. doi:10.1016/j.ajhg.2012.07.005. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22883147>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3415535/>)
- Zanotti S, Canalis E. Notch signaling in skeletal health and disease. *Eur J Endocrinol.* 2013 May 8;168(6):R95-103. doi: 10.1530/EJE-13-0115. Print 2013 Jun. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23554451>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4501254/>)

Genomic Location

The *RBPJ* gene is found on chromosome 4 (<https://medlineplus.gov/genetics/chromosome/4/>).

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