



SH3BP2 gene

SH3 domain binding protein 2

Normal Function

The *SH3BP2* gene provides instructions for making a protein whose exact function is unclear, although it is known to interact with other proteins within cells. The SH3BP2 protein plays a role in transmitting chemical signals, particularly in certain immune system cells and cells involved in the replacement of old bone tissue with new bone (bone remodeling).

Studies suggest that the SH3BP2 protein helps regulate signaling pathways that activate immune system cells called B cells and macrophages. The protein is also involved in the production of osteoclasts, which are specialized cells that break down bone tissue when it is no longer needed. Osteoclasts play a central role in bone remodeling.

Health Conditions Related to Genetic Changes

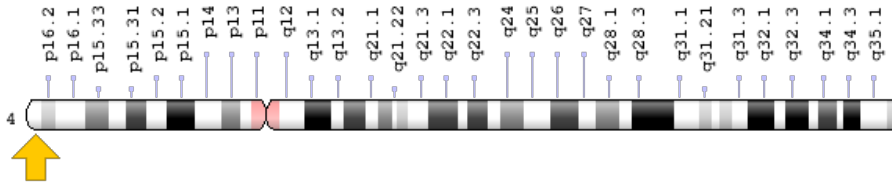
Cherubism

At least 11 mutations in the *SH3BP2* gene have been identified in people with cherubism. Each of these mutations changes a single protein building block (amino acid) in a critical region of the SH3BP2 protein. These genetic changes lead to the production of an overly active version of this protein. The effects of *SH3BP2* mutations are still under study, but researchers believe that the abnormal protein alters critical signaling pathways in cells associated with the maintenance of bone tissue and in certain immune system cells. The overactive protein likely causes inflammation in the bones of the jaw and triggers the production of an increased number of osteoclasts. An excess of these bone-eating cells contributes to the abnormal breakdown of bone tissue in the upper and lower jaws. A combination of bone loss and inflammation likely underlies the cyst-like growths characteristic of cherubism.

Chromosomal Location

Cytogenetic Location: 4p16.3, which is the short (p) arm of chromosome 4 at position 16.3

Molecular Location: base pairs 2,793,023 to 2,841,096 on chromosome 4 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 3BP-2
- 3BP2
- 3BP2_HUMAN
- CRBM
- CRPM
- FLJ42079
- RES4-23
- SH3-domain binding protein 2

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth Edition, 2002): Osteoblasts Secrete Bone Matrix, While Osteoclasts Erode It
<https://www.ncbi.nlm.nih.gov/books/NBK26889/#A4189>

Clinical Information from GeneReviews

- Cherubism
<https://www.ncbi.nlm.nih.gov/books/NBK1137>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SH3BP2%5BTIAB%5D%29+OR+%28SH3-domain+binding+protein+2%5BTIAB%5D%29%29+OR+%283BP2%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- SH3 DOMAIN-BINDING PROTEIN 2
<http://omim.org/entry/602104>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SH3BP2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SH3BP2%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10825
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:6452>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6452>
- UniProt
<https://www.uniprot.org/uniprot/P78314>

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Reviewed: April 2007
Published: June 11, 2019

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services