

## 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 turn on (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 15 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 abnormal protein that does not get broken down when it is no longer needed. Too much SH3BP2 protein likely increases signaling in certain cells, causing an immune reaction (inflammation) in the bones of the jaw and also triggering the production of an increased number of osteoclasts. An excess of these bone-destroying 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.

### Other Names for This Gene

- 3BP-2
- 3BP2
- 3BP2\_HUMAN
- CRBM

- CRPM
- FLJ42079
- RES4-23
- SH3-domain binding protein 2

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of SH3BP2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6452\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6452[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29%29%29>)

### Catalog of Genes and Diseases from OMIM

- SH3 DOMAIN-BINDING PROTEIN 2; SH3BP2 (<https://omim.org/entry/602104>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6452>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SH3BP2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SH3BP2[gene]))

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### **Genomic Location**

The *SH3BP2* gene is found on chromosome 4 (<https://medlineplus.gov/genetics/chromosome/4/>).

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