



## SALL1 gene

spalt like transcription factor 1

### Normal Function

The *SALL1* gene is part of a group of genes called the SALL family. These genes provide instructions for making proteins that are involved in the formation of tissues and organs during embryonic development. SALL proteins are transcription factors, which means they attach (bind) to specific regions of DNA and help control the activity of particular genes.

The exact function of the SALL1 protein is unclear. This protein is made in many tissues, including the kidneys, liver, and fetal and adult brain. Based on the effects of *SALL1* mutations, researchers infer that this protein plays an important role in development of the hands (particularly the thumbs), ears, anus, kidneys, and other parts of the body before birth.

### Health Conditions Related to Genetic Changes

#### Townes-Brocks Syndrome

More than 55 mutations in the *SALL1* gene have been identified in people with Townes-Brocks syndrome. Researchers originally believed that all of these mutations prevented one copy of the gene in each cell from making any protein, resulting in a shortage of SALL1 protein during development. More recently, they found that some mutations lead to the production of an abnormally small version of the SALL1 protein that malfunctions within cells. The malfunctioning protein interferes with normal copies of the SALL1 protein, preventing them from entering the nucleus to regulate gene activity. Scientists suspect that this type of genetic change likely underlies the more severe cases of Townes-Brocks syndrome. Mutations that reduce the amount of SALL1 protein are probably responsible for milder cases of this condition.

The *SALL1* gene appears to be necessary for the normal development of many different organs and tissues before birth, which helps explain why mutations in this gene can cause the varied birth defects associated with Townes-Brocks syndrome. It is uncertain, however, how *SALL1* mutations result in the specific features of this condition including an obstruction of the anal opening (imperforate anus), abnormally shaped ears, and hand malformations.

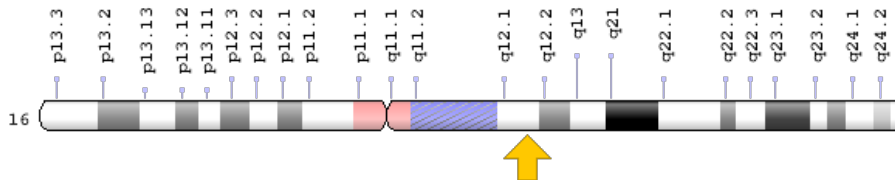
#### Coloboma

#### Congenital anomalies of kidney and urinary tract

## Chromosomal Location

Cytogenetic Location: 16q12.1, which is the long (q) arm of chromosome 16 at position 12.1

Molecular Location: base pairs 51,135,975 to 51,151,270 on chromosome 16 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- HSAL1
- sal (Drosophila)-like 1
- sal-like 1
- sal-like 1 (Drosophila)
- Sal-like protein 1
- SALL1\_HUMAN
- spalt-like transcription factor 1
- TBS
- ZNF794

## Additional Information & Resources

### Clinical Information from GeneReviews

- Townes-Brocks Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1445>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SALL1%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

- SAL-LIKE 1  
<http://omim.org/entry/602218>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SALL1.html](http://atlasgeneticsoncology.org/Genes/GC_SALL1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SALL1%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:10524](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10524)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:6299>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6299>
- UniProt  
<https://www.uniprot.org/uniprot/Q9NSC2>

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