SALL1 gene

spalt like transcription factor 1

Normal Function

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

The SALL1 protein helps turn off (repress) gene activity by interacting with other proteins that alter how tightly regions of DNA are packaged. This process, known as chromatin remodeling, is one way gene expression is regulated during development; typically, when DNA is tightly packed, gene expression is lower than when DNA is loosely packed. By controlling gene activity, the SALL1 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

At least 75 mutations in the *SALL1* gene have been identified in people with Townes-Brocks syndrome, which is typically characterized by a malformation of the anal opening (imperforate anus), abnormally shaped ears, and hand malformations. It is uncertain how these mutations result in the specific features of the condition.

Most *SALL1* gene mutations involved in this disorder lead to the production of an abnormally short version of the SALL1 protein that malfunctions within cells. The malfunctioning protein is thought to interfere with normal copies of the SALL1 protein, which are produced from the other copy of the *SALL1* gene that does not have a mutation. This interference prevents the normal proteins from entering the nucleus to regulate gene activity. The malfunctioning protein may also interact with other proteins, disrupting their function. For example, some research indicates that the abnormally short SALL1 protein interferes with proteins that control the formation of cellular structures called cilia. Cilia are microscopic, finger-like projections that stick out from the surface of cells and participate in signaling pathways that transmit information within and between cells. Cilia are important for the structure and function of many types of cells and the normal development of several tissues. Abnormalities in cilia can disrupt

important chemical signaling pathways during development and may contribute to the features of Townes-Brocks syndrome.

Other, less-common *SALL1* gene mutations prevent the gene from making protein; this reduces by half the amount of SALL1 protein produced in cells. Some studies suggest that mutations that reduce the amount of SALL1 protein are responsible for milder cases of Townes-Brocks syndrome, and mutations that result in an abnormally short, malfunctioning SALL1 protein underlie the more severe cases of the condition. However, other studies do not find the same correlation.

A shortage of functioning SALL1 protein, due to either type of mutation, likely impairs the regulation of genes that direct the development of many different organs and tissues before birth. Abnormal development before birth due to unusual gene activity leads to the varied birth defects associated with Townes-Brocks syndrome.

Coloboma

MedlinePlus Genetics provides information about Coloboma

Congenital anomalies of kidney and urinary tract

MedlinePlus Genetics provides information about Congenital anomalies of kidney and urinary tract

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

Tests Listed in the Genetic Testing Registry

- Tests of SALL1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6299[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; SALL1 (https://omim.org/entry/602218)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6299)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SALL1[gene])

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Genomic Location

The *SALL1* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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