SALL4 gene

spalt like transcription factor 4

Normal Function

The *SALL4* 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 SALL4 protein remains unclear. Based on the functions of similar proteins in other organisms (such as zebrafish and mice), the SALL4 protein appears to play a critical role in the developing limbs. This protein may also be important for the development of nerves that control eye movement and for the formation of the walls (septa) that divide the heart into separate chambers.

Health Conditions Related to Genetic Changes

Duane-radial ray syndrome

More than 25 mutations in the *SALL4* gene have been identified in people with Duane-radial ray syndrome (also known as Okihiro syndrome) or a very similar condition called acro-renal-ocular syndrome. Researchers suspect that Duane-radial ray syndrome and acro-renal-ocular syndrome are part of an overlapping set of syndromes with many possible signs and symptoms.

Most *SALL4* gene mutations create a premature stop signal in the instructions for making the SALL4 protein. As a result, cells do not produce any functional protein from one copy of this gene. Researchers are investigating how a reduction in the amount of the SALL4 protein disrupts eye, heart, and limb development in people with Duane-radial ray syndrome and acro-renal-ocular syndrome.

Anophthalmia/Microphthalmia

MedlinePlus Genetics provides information about Anophthalmia/Microphthalmia

Coloboma

MedlinePlus Genetics provides information about Coloboma

Other disorders

A least one mutation in the *SALL4* gene has been found to cause IVIC syndrome, a condition whose signs and symptoms overlap with those of Duane-radial ray syndrome and acro-renal-ocular syndrome (described above). The acronym IVIC stands for Instituto Venezolano de Investigaciones Cientìficas, the center where the condition was first described. Major features of IVIC syndrome include abnormally formed bones in the arms and hands, hearing loss, and problems with eye movement caused by abnormalities of the muscles that surround the eyes (extraocular muscles).

The *SALL4* gene mutation responsible for IVIC syndrome creates a premature stop signal in the instructions for making the SALL4 protein. As a result, cells produce an abnormally short version of the protein from one copy of this gene. It is unclear whether this shortened protein is completely nonfunctional or if it retains some of its function as a transcription factor. Researchers are working to determine how this *SALL4* gene mutation disrupts early development and leads to the characteristic features of IVIC syndrome.

Other Names for This Gene

- dJ1112F19.1
- DRRS
- HSAL4
- MGC133050
- sal (Drosophila)-like 4
- sal-like 4
- sal-like 4 (Drosophila)
- SALL4_HUMAN
- spalt-like transcription factor 4
- Zinc finger protein SALL4
- ZNF797

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SALL4 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57167[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SALL4%5BTIAB%5D%29+OR+%28sal-like+4%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+A

ND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- IVIC SYNDROME; IVIC (https://omim.org/entry/147750)
- SAL-LIKE 4; SALL4 (https://omim.org/entry/607343)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/57167)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SALL4[gene])

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Genomic Location

The *SALL4* gene is found on chromosome 20 (https://medlineplus.gov/genetics/chromosome/20/).

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