



## SOX9 gene

SRY-box 9

### Normal Function

The *SOX9* gene provides instructions for making a protein that plays a critical role during embryonic development. The *SOX9* protein is especially important for development of the skeleton and plays a key role in the determination of sex before birth. The *SOX9* protein attaches (binds) to specific regions of DNA and regulates the activity of other genes, particularly those that control skeletal development and sex determination. On the basis of this action, the *SOX9* protein is called a transcription factor.

### Health Conditions Related to Genetic Changes

#### Campomelic dysplasia

More than 70 mutations involving the *SOX9* gene have been found to cause campomelic dysplasia, a disorder that affects skeletal development, sex determination, and other processes in the body and is often life-threatening in the newborn period. Most cases of campomelic dysplasia are caused by mutations within the *SOX9* gene. These mutations prevent the production of the *SOX9* protein or result in a protein with impaired ability to function as a transcription factor. About 5 percent of cases are caused by chromosome abnormalities that occur near the *SOX9* gene. These chromosome abnormalities disrupt regions of DNA called enhancers that normally regulate the activity of the *SOX9* gene. All of these genetic changes prevent the *SOX9* protein from properly controlling the genes essential for normal development of the skeleton, reproductive organs, and other parts of the body. Abnormal development of these structures causes the signs and symptoms of campomelic dysplasia.

Individuals with milder forms of campomelic dysplasia are more likely to have chromosome abnormalities near the *SOX9* gene rather than mutations within the gene.

#### Isolated Pierre Robin sequence

Genetic changes that occur near the *SOX9* gene cause some cases of isolated Pierre Robin sequence. Individuals with this condition have a small lower jaw (micrognathia) and a tongue that is placed further back than normal (glossoptosis), which can block the airways. Most affected individuals are also born with an opening in the roof of the mouth (a cleft palate). These cases of Pierre Robin sequence are described as isolated because they occur without other signs and symptoms.

The genetic changes associated with isolated Pierre Robin sequence are thought to disrupt enhancer regions that normally regulate the activity of the SOX9 gene during development of the lower jaw, which reduces SOX9 gene activity. As a result, the SOX9 protein cannot properly control the genes essential for normal jaw development, causing micrognathia. Underdevelopment of the lower jaw affects placement of the tongue and formation of the palate, leading to glossoptosis and, often, cleft palate.

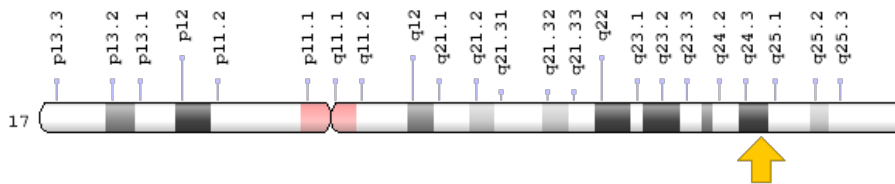
46,XX testicular disorder of sex development

Swyer syndrome

### Chromosomal Location

Cytogenetic Location: 17q24.3, which is the long (q) arm of chromosome 17 at position 24.3

Molecular Location: base pairs 72,121,020 to 72,126,420 on chromosome 17 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- SOX9\_HUMAN
- SRA1
- SRY (sex determining region Y)-box 9
- SRY (sex-determining region Y)-box 9 protein
- SRY box 9
- transcription factor SOX9

## **Additional Information & Resources**

### Educational Resources

- Developmental Biology (sixth edition, 2000): Sox9: Autosomal Sex Reversal  
<https://www.ncbi.nlm.nih.gov/books/NBK9967/#A4117>
- Transcriptional Control of Neural Crest Development (2010): Sox9  
<https://www.ncbi.nlm.nih.gov/books/NBK53136/#s13.5>

### Clinical Information from GeneReviews

- Campomelic Dysplasia  
<https://www.ncbi.nlm.nih.gov/books/NBK1760>
- Nonsyndromic 46,XX Testicular Disorders of Sex Development  
<https://www.ncbi.nlm.nih.gov/books/NBK1416>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SOX9%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- SRY-BOX 9  
<http://omim.org/entry/608160>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SOX9.html](http://atlasgeneticsoncology.org/Genes/GC_SOX9.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SOX9%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:11204](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:11204)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:6662>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6662>
- UniProt  
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<https://ghr.nlm.nih.gov/gene/SOX9>

Reviewed: December 2016  
Published: May 14, 2019

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services