

## SOX11 gene

SRY-box transcription factor 11

### Normal Function

The *SOX11* gene provides instructions for making a protein that plays a critical role in the development of the brain and nerve cells (neurons). This protein is a transcription factor, which means it attaches (binds) to specific regions of DNA and coordinates with other proteins to turn on particular genes.

Studies suggest that the SOX11 protein may also be able to regulate the activity of genes involved in brain and nerve cell development through a different process known as chromatin remodeling. Chromatin is the network of DNA and protein that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly regions of DNA are packaged. Chromatin remodeling is one way gene expression is regulated during development; when DNA is tightly packed, gene expression is often lower than when DNA is loosely packed.

The activity (expression) of the *SOX11* gene is controlled by chromatin remodeling by special protein groups called SWI/SNF complexes.

### Health Conditions Related to Genetic Changes

#### Coffin-Siris syndrome

At least 14 variants (also known as mutations) in the *SOX11* gene have been found to cause Coffin-Siris syndrome. This condition is characterized by intellectual disability, abnormalities of the fifth (pinky) fingers or toes, and characteristic facial features that are described as coarse. Most *SOX11* gene variants involved in Coffin-Siris syndrome alter the SOX11 protein or prevent its production. These changes reduce the ability of the SOX11 protein to regulate the activity of genes, including those important for development of the brain and nerve cells. Altered gene activity can disrupt development of the brain and other tissues and organs, which could explain the diverse signs and symptoms of Coffin-Siris syndrome.

### Other Names for This Gene

- SRY (sex determining region Y)-box 11
- SRY-box 11

- SRY-related HMG-box gene 11

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of SOX11 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6664\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6664[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=SOX11>)

### Catalog of Genes and Diseases from OMIM

- SRY-BOX 11; SOX11 (<https://omim.org/entry/600898>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6664>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SOX11\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SOX11[gene]))

## **References**

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