SOX2 gene

SRY-box transcription factor 2

Normal Function

The *SOX2* gene provides instructions for making a protein that plays a critical role in the formation of many different tissues and organs during embryonic development. The SOX2 protein is especially important for the development of the eyes. This protein regulates the activity of other genes by attaching (binding) to specific regions of DNA. On the basis of this action, the SOX2 protein is called a transcription factor.

Health Conditions Related to Genetic Changes

Septo-optic dysplasia

MedlinePlus Genetics provides information about Septo-optic dysplasia

SOX2 anophthalmia syndrome

At least 33 mutations in the *SOX2* gene have been found to cause *SOX2* anophthalmia syndrome. Some of these mutations prevent the gene from making any SOX2 protein, while others result in the production of an abnormally short, nonfunctional version of the protein. A few mutations change single protein building blocks (amino acids) in the SOX2 protein. All of these mutations disrupt the protein's ability to regulate genes essential for normal development of the eyes and other parts of the body. Abnormal development of these structures causes the signs and symptoms of *SOX2* anophthalmia syndrome.

Coloboma

MedlinePlus Genetics provides information about Coloboma

Combined pituitary hormone deficiency

MedlinePlus Genetics provides information about Combined pituitary hormone deficiency

Microphthalmia

MedlinePlus Genetics provides information about Microphthalmia

Other Names for This Gene

- ANOP3
- MCOPS3
- MGC2413
- sex-determining region Y-box 2
- SOX2_HUMAN
- SRY (sex determining region Y)-box 2
- SRY box 2
- SRY-related HMG-box gene 2
- transcription factor SOX2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SOX2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6657[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SOX2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28stem+cell%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SRY-BOX 2; SOX2 (https://omim.org/entry/184429)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6657)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SOX2[gene])

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Genomic Location

The *SOX2* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromoso

me/3/).

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