SOX2 anophthalmia syndrome

Description

SOX2 anophthalmia syndrome is a rare disorder characterized by abnormal development of the eyes and other parts of the body.

People with *SOX2* anophthalmia syndrome are usually born without eyeballs (anophthalmia), although some individuals have small eyes (microphthalmia). The term anophthalmia is often used interchangeably with severe microphthalmia because individuals with no visible eyeballs typically have some remaining eye tissue. These eye problems can cause significant vision loss. While both eyes are usually affected in *SOX2* anophthalmia syndrome, one eye may be more affected than the other.

Individuals with *SOX2* anophthalmia syndrome may also have seizures, brain abnormalities, slow growth, delayed development of motor skills (such as walking), and mild to severe learning disabilities. Some people with this condition are born with a blocked esophagus (esophageal atresia), which is often accompanied by an abnormal connection between the esophagus and the trachea (tracheoesophageal fistula). Genital abnormalities have been described in affected individuals, especially males. Male genital abnormalities include undescended testes (cryptorchidism) and an unusually small penis (micropenis).

Frequency

SOX2 anophthalmia syndrome is estimated to affect 1 in 250,000 individuals. About 10 percent to 15 percent of people with anophthalmia in both eyes have *SOX2* anophthalmia syndrome.

Causes

Mutations in the *SOX2* gene cause *SOX2* anophthalmia syndrome. This 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 regulates the activity of other genes, especially those that are important for normal development of the eyes.

Mutations in the *SOX2* gene prevent the production of functional SOX2 protein. The absence of this protein disrupts the activity of genes that are essential for the development of the eyes and other parts of the body. Abnormal development of these

structures causes the signs and symptoms of *SOX2* anophthalmia syndrome.

Learn more about the gene associated with SOX2 anophthalmia syndrome

- SOX2

Inheritance

SOX2 anophthalmia syndrome is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Most cases result from new mutations in the *SOX2* gene and occur in people with no history of the disorder in their family. In a small number of cases, people with *SOX2* anophthalmia syndrome have inherited the altered gene from an unaffected parent who has a *SOX2* mutation only in their sperm or egg cells. This phenomenon is called germline mosaicism.

Other Names for This Condition

- AEG syndrome
- Anophthalmia-esophageal-genital syndrome
- SOX2-related eye disorders
- Syndromic microphthalmia 3

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Anophthalmia/microphthalmia-esophageal atresia syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1859773/)

Genetic and Rare Diseases Information Center

- Anophthalmia/microphthalmia-esophageal atresia syndrome (https://rarediseases.info.nih.gov/diseases/1443/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- MICROPHTHALMIA, SYNDROMIC 3; MCOPS3 (https://omim.org/entry/206900)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SOX2+anophthalmia+syndrome%5BTIAB%5D%29+OR+%28SOX2%5BTIAB%5D%29%29+AND+%28eye%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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