OTX2 gene

orthodenticle homeobox 2

Normal Function

The *OTX2* gene provides instructions for producing a protein that regulates the activity of other genes. On the basis of this action, the OTX2 protein is called a transcription factor. The *OTX2* gene is part of a family of homeobox genes, which act during early embryonic development to control the formation of many body structures.

The *OTX2* gene plays a critical role in the development of the eyes and related structures, such as the nerves that carry visual information from the eyes to the brain (optic nerves). It is also involved in brain development, including the formation of the pituitary gland at the base of the brain. The pituitary gland produces hormones that help control growth, reproduction, and other critical body functions.

Health Conditions Related to Genetic Changes

Septo-optic dysplasia

At least eight mutations in the *OTX2* gene have been identified in people with the major features of septo-optic dysplasia. Some of these mutations prevent the production of a functional OTX2 protein. Other mutations lead to a defective version of the protein that cannot regulate the activity of other genes.

A shortage of the OTX2 protein disrupts the formation and early development of the eyes, the optic nerves, the pituitary gland, and other brain structures. These problems with development lead to the major features of septo-optic dysplasia, including eye abnormalities, underdevelopment of the pituitary gland (pituitary hypoplasia), and learning difficulties. However, the signs and symptoms associated with *OTX2* gene mutations vary widely, even among affected members of the same family. Additional features that have been reported in people with *OTX2* gene mutations include delayed development, slow growth, and seizures.

Studies suggest that mutations in the *OTX2* gene are a rare cause of septo-optic dysplasia.

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

- homeobox protein OTX2
- MCOPS5
- MGC45000
- orthodenticle homolog 2
- OTX2_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of OTX2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5015[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28OTX2%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ORTHODENTICLE HOMEOBOX 2; OTX2 (https://omim.org/entry/600037)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5015)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=OTX2[gene])

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Genomic Location

The *OTX2* gene is found on chromosome 14 (https://medlineplus.gov/genetics/chromosome/14/).

Last updated March 1, 2010