Lenz microphthalmia syndrome

Description

Lenz microphthalmia syndrome is a condition characterized by abnormal development of the eyes and several other parts of the body. It occurs almost exclusively in males.

The eye abnormalities associated with Lenz microphthalmia syndrome can affect one or both eyes. People with this condition are born with eyeballs that are abnormally small (microphthalmia) or absent (anophthalmia), leading to vision loss or blindness. Other eye problems can include clouding of the lens (cataract), involuntary eye movements (nystagmus), a gap or split in structures that make up the eye (coloboma), and a higher risk of an eye disease called glaucoma.

Abnormalities of the ears, teeth, hands, skeleton, and urinary system are also frequently seen in Lenz microphthalmia syndrome. Less commonly, heart defects have been reported in affected individuals. Many people with this condition have delayed development or intellectual disability ranging from mild to severe.

Frequency

Lenz microphthalmia syndrome is a very rare condition; its incidence is unknown. It has been identified in only a few families worldwide.

Causes

Mutations in at least two genes on the X chromosome are thought to be responsible for Lenz microphthalmia syndrome. Only one of these genes, *BCOR*, has been identified.

The *BCOR* gene provides instructions for making a protein called the BCL6 corepressor. This protein helps regulate the activity of other genes. Little is known about the protein' s function, although it appears to play an important role in early embryonic development. A mutation in the *BCOR* gene has been found in one family with Lenz microphthalmia syndrome. This mutation changes the structure of the BCL6 corepressor protein, which disrupts the normal development of the eyes and several other organs and tissues before birth.

Researchers are working to determine whether Lenz microphthalmia syndrome is a single disorder with different genetic causes or two very similar disorders, each caused by mutations in a different gene. They are searching for a second gene on the X

chromosome that may underlie additional cases of the disorder.

Learn more about the gene associated with Lenz microphthalmia syndrome

- BCOR

Inheritance

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

Other Names for This Condition

- Lenz dysmorphogenic syndrome
- Lenz dysplasia
- Lenz syndrome
- MAA
- MCOPS1
- Microphthalmia or anophthalmos with associated anomalies
- Microphthalmia, syndromic 1

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Lenz microphthalmia syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0796016/)

Genetic and Rare Diseases Information Center

- Lenz microphthalmia syndrome (https://rarediseases.info.nih.gov/diseases/87/lenz-microphthalmia-syndrome)

Patient Support and Advocacy Resources

- Disease InfoSearch (https://www.diseaseinfosearch.org/)

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

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov (https://clinicaltrials.gov/ct2/results?cond=%22Eye+Diseases%2C+Hereditary%22+OR+%22Lenz+microphthalmia+syndrome%22)

Catalog of Genes and Diseases from OMIM

- MICROPHTHALMIA, SYNDROMIC 1 (https://omim.org/entry/309800)

Scientific Articles on PubMed

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

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