Oculofaciocardiodental syndrome

Description

Oculofaciocardiodental (OFCD) syndrome is a condition that affects the development of the eyes (oculo-), facial features (facio-), heart (cardio-), and teeth (dental).

The eye abnormalities associated with OFCD syndrome can affect one or both eyes. Many people with this condition are born with eyeballs that are abnormally small (microphthalmia). Other eye problems can include clouding of the lens (cataract) and a high risk of glaucoma, an eye disease that increases the pressure in the eye. These abnormalities can lead to vision loss or blindness.

People with OFCD syndrome often have a long, narrow face with distinctive facial features, including deep-set eyes, droopy eyelids (ptosis), and a nose with a high bridge and broad tip. Affected individuals may have a split (cleft) in their nose or in the roof of their mouth (cleft palate).

Heart defects are another common feature of OFCD syndrome. Babies with this condition may be born with a hole between two chambers of the heart (an atrial or ventricular septal defect) or a leak in one of the valves that controls blood flow through the heart (mitral valve prolapse).

Teeth with very large roots (radiculomegaly) are characteristic of OFCD syndrome. Additional dental abnormalities can include the delayed loss of primary (baby) teeth, missing or abnormally small teeth, delayed teething (dentition), misaligned teeth, and defective tooth enamel.

Individuals with OFCD syndrome can have additional features, such as skeletal abnormalities (typically affecting the toes), hearing loss, and intellectual disabilities.

Frequency

OFCD syndrome is very rare; the incidence is estimated to be less than 1 in 1 million people.

Causes

Variants (also called mutations) in the *BCOR* gene cause OFCD syndrome. The *BCOR* gene provides instructions for making a protein called the BCL6 corepressor. This

protein helps regulate the activity of other genes. Specifically, the BCL6 corepressor appears to play an important role in regulating genes during early development, particularly those that are involved in the formation of the eye and other organs and tissues.

The variants in the *BCOR* gene that cause OFCD syndrome prevent the production of any functional BCL6 corepressor protein. As a result, gene regulation during development is disrupted, which impairs the normal development of the eyes and several other organs and tissues before birth.

Learn more about the gene associated with Oculofaciocardiodental syndrome

- BCOR

Inheritance

OFCD syndrome is inherited in an X-linked dominant pattern. The *BCOR* gene is located on the X chromosome, which is one of the two sex chromosomes. In females (who have two X chromosomes), a variant in one of the two copies of the gene in each cell is sufficient to cause the disorder. As a result, some cells produce a normal amount of BCL6 corepressor protein and other cells produce none, leading to about half the normal amount of protein.

No males (with only one X chromosome) have been born with OFCD syndrome. A BCOR gene variant in the only copy of the gene would cause a complete lack of BCL6 corepressor protein, and this is thought to be lethal very early in development.

Other Names for This Condition

- MCOPS2
- Microphthalmia, cataracts, radiculomegaly, and septal heart defects
- Microphthalmia, syndromic 2
- Oculo-facio-cardio-dental syndrome
- OFCD syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Oculofaciocardiodental syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1846265/)

Genetic and Rare Diseases Information Center

- Oculofaciocardiodental syndrome (https://rarediseases.info.nih.gov/diseases/4628/i

ndex)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- MICROPHTHALMIA, SYNDROMIC 2; MCOPS2 (https://omim.org/entry/300166)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28oculofaciocardiodental+syndrome%5BTIAB%5D%29+OR+%28mcops2%5BTIAB%5D%29+OR+%28ofcd+syndrome%5BTIAB%5D%29+OR+%28oculo-facio-cardio-dental+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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