Blepharocheilodontic syndrome

Description

Blepharocheilodontic (BCD) syndrome is a disorder that is present at birth. It mainly affects the eyelids (blepharo-), upper lip (-cheilo-), and teeth (-dontic).

People with BCD syndrome have lower eyelids that turn out so that the inner surface is exposed (ectropion). The outside of the lower lid may sag away from the eye (euryblepharon), and the eyelids may not be able to close completely (lagophthalmia). There can be extra eyelashes (distichiasis) on the upper eyelids, ranging from a few extra eyelashes to a full extra set. These eyelashes do not grow along the edge of the eyelid with the normal lashes, but out of its inner lining. When the abnormal eyelashes touch the eyeball, they can cause damage to the clear covering of the eye (cornea). Affected individuals may also have widely spaced eyes (hypertelorism), a flat face, and a high forehead.

Other features of BCD syndrome usually include openings on both sides of the upper lip (bilateral cleft lip) and an opening in the roof of the mouth (cleft palate). Affected individuals may have fewer teeth than normal (oligodontia) and their teeth are often smaller than usual and cone-shaped. The dental abnormalities affect both primary teeth (sometimes called "baby teeth") and secondary (permanent) teeth. Other frequent features include sparse, fine hair and abnormal nails.

Occasionally people with BCD syndrome have additional features, including an obstruction of the anal opening (imperforate anus); malformation or absence of the butterfly-shaped gland in the lower neck called the thyroid, resulting in lack of thyroid gland function; or fused fingers or toes (syndactyly). Very rarely, affected individuals have incompletely formed arms or legs (limb reduction defects) or a spinal cord abnormality known as spina bifida.

Frequency

BCD syndrome is a rare disorder; its prevalence is unknown. At least 50 affected individuals have been described in the medical literature.

Causes

BCD syndrome is caused by mutations in the *CDH1* or *CTNND1* gene. These genes provide instructions for making proteins called epithelial cadherin (E-cadherin) and p120-

catenin, respectively. The E-cadherin protein is found within the membrane that surrounds epithelial cells, which are the cells that line the surfaces and cavities of the body, including the inside of the eyelids and mouth. This protein is involved in the attachment of cells to one another (cell adhesion). The p120-catenin protein helps keep E-cadherin in its proper place in the cell membrane, preventing it from being taken into the cell and broken down prematurely. Interactions between the two proteins are also important for other cell processes that are involved in the development of the head and face (craniofacial development), including the eyelids and teeth.

While the specific effects of the *CDH1* gene mutations that cause BCD syndrome are not well understood, researchers suggest that the mutations may result in an E-cadherin protein that is unstable and quickly broken down. *CTNND1* gene mutations reduce or eliminate the production or function of p120-catenin, so E-cadherin is broken down prematurely. A shortage of these proteins disrupts normal development, especially craniofacial development, which is thought to underlie the features of BCD syndrome.

Learn more about the genes associated with Blepharocheilodontic syndrome

- CDH1
- CTNND1

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In some cases, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

Other Names for This Condition

- BCD syndrome
- BCDS
- Blepharo-cheilo-dontic syndrome
- Blepharo-cheilo-odontic syndrome
- Clefting, ectropion, and conical teeth
- Ectropion, inferior, with cleft lip and/or palate
- Elschnig syndrome
- Lagophthalmia with bilateral cleft lip and palate

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Blepharocheilodontic syndrome 1 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4551988/)
- Genetic Testing Registry: Blepharocheilodontic syndrome 2 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4540127/)

Genetic and Rare Diseases Information Center

- Blepharo-cheilo-odontic syndrome (https://rarediseases.info.nih.gov/diseases/2071/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- BLEPHAROCHEILODONTIC SYNDROME 1; BCDS1 (https://omim.org/entry/119580)
- BLEPHAROCHEILODONTIC SYNDROME 2; BCDS2 (https://omim.org/entry/617681)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28blepharo-cheilo-dontic+syndrome%5BTIAB%5D%29+OR+%28blepharocheilodontic+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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