

Oculodentodigital dysplasia

Description

Oculodentodigital dysplasia is a condition that affects many parts of the body, particularly the eyes (oculo-), teeth (dento-), and fingers (digital). Common features in people with this condition are small eyes (microphthalmia) and other eye abnormalities that can lead to vision loss. Affected individuals also frequently have tooth abnormalities, such as small or missing teeth, weak enamel, multiple cavities, and early tooth loss. Other common features of this condition include a thin nose and webbing of the skin (syndactyly) between the fourth and fifth fingers.

Less common features of oculodentodigital dysplasia include sparse hair growth (hypotrichosis), brittle nails, an unusual curvature of the fingers (camptodactyly), syndactyly of the toes, small head size (microcephaly), and an opening in the roof of the mouth (cleft palate). Some affected individuals experience neurological problems such as a lack of bladder or bowel control, difficulty coordinating movements (ataxia), abnormal muscle stiffness (spasticity), hearing loss, and impaired speech (dysarthria). A few people with oculodentodigital dysplasia also have a skin condition called palmoplantar keratoderma. Palmoplantar keratoderma causes the skin on the palms and the soles of the feet to become thick, scaly, and calloused.

Some features of oculodentodigital dysplasia are evident at birth, while others become apparent with age.

Frequency

The exact incidence of oculodentodigital dysplasia is unknown. It has been diagnosed in fewer than 1,000 people worldwide. More cases are likely undiagnosed.

Causes

Mutations in the *GJA1* gene cause oculodentodigital dysplasia. The *GJA1* gene provides instructions for making a protein called connexin 43. This protein forms one part (a subunit) of channels called gap junctions, which allow direct communication between cells. Gap junctions formed by connexin 43 proteins are found in many tissues throughout the body.

GJA1 gene mutations result in abnormal connexin 43 proteins. Channels formed with abnormal proteins are often permanently closed. Some mutations prevent connexin 43

proteins from traveling to the cell surface where they are needed to form channels between cells. Impaired functioning of these channels disrupts cell-to-cell communication, which likely interferes with normal cell growth and cell specialization, processes that determine the shape and function of many different parts of the body. These developmental problems cause the signs and symptoms of oculodentodigital dysplasia.

[Learn more about the gene associated with Oculodentodigital dysplasia](#)

- GJA1

Inheritance

Most cases of oculodentodigital dysplasia are 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.

Less commonly, oculodentodigital dysplasia can be inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition. Fewer than ten cases of autosomal recessive oculodentodigital dysplasia have been reported.

Other Names for This Condition

- Oculo-dento-digital dysplasia
- Oculo-dento-osseous dysplasia
- Oculodentodigital syndrome
- Oculodentoosseous dysplasia
- ODD syndrome
- ODDD
- ODOD
- Osseous-oculo-dental dysplasia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Oculodentodigital dysplasia (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0812437/>)

Genetic and Rare Diseases Information Center

- Oculodentodigital dysplasia (<https://rarediseases.info.nih.gov/diseases/7239/index>)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- OCULODENTODIGITAL DYSPLASIA; ODDD (<https://omim.org/entry/164200>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28oculodentodigital+dysplasia%5BTIAB%5D%29+OR+%28oculo-dento-digital+dysplasia%5BTIAB%5D%29+OR+%28oddd%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>)

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