

Distal 18q deletion syndrome

Description

Distal 18q deletion syndrome is a chromosomal condition that occurs when a piece of the long (q) arm of chromosome 18 is missing. The term "distal" means that the missing piece occurs near one end of the chromosome. Distal 18q deletion syndrome can lead to a wide variety of signs and symptoms among affected individuals.

Some common features of distal 18q deletion syndrome include short stature (often due to growth hormone deficiency), weak muscle tone (hypotonia), hearing loss due to ear canals that are narrow (aural stenosis) or absent (aural atresia), and foot abnormalities such as an inward or upward-turning foot (clubfoot) or feet with soles that are rounded outward (rocker-bottom feet). Eye movement disorders and other vision problems, an opening in the roof of the mouth (cleft palate), an underactive thyroid gland (hypothyroidism), heart abnormalities that are present from birth (congenital heart defects), kidney problems, genital abnormalities, and skin problems may also occur in this disorder. Some affected individuals have mild facial differences such as deep-set eyes, a flat or sunken appearance of the middle of the face (midface hypoplasia), a wide mouth, and prominent ears. These features are often not noticeable except in a detailed medical evaluation.

Distal 18q deletion syndrome can also affect the nervous system. A common neurological feature of this disorder is impaired myelin production (dysmyelination). Myelin is a fatty substance that insulates nerve cells and promotes the rapid transmission of nerve impulses. The formation of a protective myelin sheath around nerve cells (myelination) normally begins before birth and continues into adulthood. In people with distal 18q deletion syndrome, myelination is often delayed and proceeds more slowly than normal; affected individuals may never have normal adult myelin levels. Most people with distal 18q deletion syndrome have neurological problems, although it is unclear to what extent these problems are related to the dysmyelination. These problems include delayed development, learning disabilities, and intellectual disability that can range from mild to severe. Seizures; hyperactivity; mood disorders such as anxiety, irritability, and depression; and features of autism spectrum disorder that affect communication and social interaction may also occur. Some affected individuals have an unusually small head size (microcephaly).

Frequency

Deletions from the q arm of chromosome 18 occur in an estimated 1 in 55,000 newborns worldwide. Most of these deletions occur in the distal region of the q arm, leading to distal 18q deletion syndrome.

Causes

Distal 18q deletion syndrome is caused by a deletion of genetic material from one copy of chromosome 18 anywhere between a region called 18q21 and the end of the chromosome. The size of the deletion and where it begins vary among affected individuals. The signs and symptoms of distal 18q deletion syndrome are thought to be related to the loss of multiple genes, some of which have not been identified, from this part of chromosome 18. Certain features of the disorder have been associated with the loss of particular genes in this region. People with deletions that include the *TCF4* gene usually have signs and symptoms of another genetic condition known as Pitt-Hopkins syndrome, such as severe intellectual disability and breathing problems, in addition to other features of distal 18q deletion syndrome.

[Learn more about the gene and chromosome associated with Distal 18q deletion syndrome](#)

- TCF4
- chromosome 18

Additional Information from NCBI Gene:

- TSHZ1

Inheritance

Distal 18q deletion syndrome is considered to be an autosomal dominant condition, which means one copy of the deleted region on chromosome 18 in each cell is sufficient to cause the disorder's characteristic features.

Most cases of distal 18q deletion syndrome are the result of a new (de novo) deletion and are not inherited. The deletion occurs most often as a random event during the formation of reproductive cells (eggs or sperm) or in early fetal development. Affected people typically have no history of the disorder in their family.

In some cases, distal 18q deletion syndrome is inherited, usually from an affected parent with relatively mild signs and symptoms. The condition can also be inherited from an unaffected parent who carries a chromosomal rearrangement called a balanced translocation, in which no genetic material is gained or lost. Individuals with a balanced translocation do not usually have any related health problems; however, the translocation can become unbalanced as it is passed to the next generation. Children

who inherit an unbalanced translocation can have a chromosomal rearrangement with extra or missing genetic material. Inheritance of an unbalanced translocation that results in the deletion of genetic material from the distal region of the q arm of chromosome 18 causes distal 18q deletion syndrome.

Other Names for This Condition

- 18q deletion syndrome
- 18q- syndrome
- Chromosome 18 long arm deletion syndrome
- Chromosome 18q deletion syndrome
- Chromosome 18q monosomy
- Chromosome 18q- syndrome
- Del(18q) syndrome
- Monosomy 18q

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Deletion of long arm of chromosome 18 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0432443/>)

Genetic and Rare Diseases Information Center

- Monosomy 18q (<https://rarediseases.info.nih.gov/diseases/10865/index>)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- CHROMOSOME 18q DELETION SYNDROME (<https://omim.org/entry/601808>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%2818q+deletion+syndrome%5BTIAB%5D%29+OR+%28monosomy+18q%5BTIAB%5D%29+OR+%2818q+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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