Proximal 18q deletion syndrome

Description

Proximal 18q deletion syndrome is a chromosomal condition that occurs when a piece of the long (q) arm of chromosome 18 is missing. The term "proximal" means that the missing piece occurs near the center of the chromosome. Individuals with proximal 18q deletion syndrome have a wide variety of signs and symptoms. Because only a small number of people are known to have this type of deletion, it can be difficult to determine which features should be considered characteristic of the disorder.

Most people with proximal 18q deletion syndrome have delayed development of skills such as sitting, crawling, walking, and speaking, and intellectual disability that can range from mild to severe. In particular, vocabulary and the production of speech (expressive language skills) may be delayed. Recurrent seizures (epilepsy) and weak muscle tone (hypotonia) often occur in this disorder. Affected individuals also frequently have neurodevelopmental disorders such as hyperactivity, aggression, and autism spectrum disorder that affect communication and social interaction.

Frequency

Deletions from the q arm of chromosome 18 occur in an estimated 1 in 55,000 newborns worldwide. However, only a small number of these individuals have deletions in the region associated with proximal 18q deletion syndrome. At least 15 people with proximal 18q deletion syndrome have been described in the medical literature.

Causes

Proximal 18q deletion syndrome is caused by a deletion of genetic material from one copy of chromosome 18. The deletion occurs near the middle of the q arm of the chromosome, typically in an area between regions called 18q11.2 and 18q21.2. The size of the deletion varies among affected individuals. The signs and symptoms of proximal 18q deletion syndrome are thought to be related to the loss of multiple genes from this part of chromosome 18. Researchers are working to determine how the loss of specific genes in this region contributes to the various features of this disorder.

Learn more about the chromosome associated with Proximal 18q deletion syndrome

- chromosome 18

Inheritance

Proximal 18q deletion syndrome is considered to be an autosomal dominant condition. This means that a deletion in one of the two copies of chromosome 18 in each cell is sufficient to cause the disorder's characteristic features.

Most cases of proximal 18q deletion syndrome are the result of a new (de novo) deletion and are not inherited from a parent. 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.

Other Names for This Condition

- 18q deletion syndrome
- 18q- syndrome
- Chromosome 18 deletion syndrome
- Chromosome 18 long arm 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

- Proximal chromosome 18q deletion syndrome (https://rarediseases.info.nih.gov/diseases/10866/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=%28proximal+18q+deletion+syndrome%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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