22q11.2 duplication

Description

22q11.2 duplication is a condition caused by an extra copy of a small piece of chromosome 22. The duplication occurs near the middle of the chromosome at a location designated q11.2.

The features of this condition vary widely, even among members of the same family. Affected individuals may have developmental delay, intellectual disability, slow growth leading to short stature, and weak muscle tone (hypotonia). Many people with the duplication have no apparent physical or intellectual disabilities.

Frequency

The prevalence of the 22q11.2 duplication in the general population is difficult to determine. Because many individuals with this duplication have no associated symptoms, their duplication may never be detected.

Most people tested for the 22q11.2 duplication have come to medical attention as a result of developmental delay or other problems affecting themselves or a family member. In one study, about 1 in 700 people tested for these reasons had the 22q11.2 duplication. Overall, more than 60 individuals with the duplication have been identified.

Causes

People with 22q11.2 duplication have an extra copy of some genetic material at position q11.2 on chromosome 22. In most cases, this extra genetic material consists of a sequence of about 3 million DNA building blocks (base pairs), also written as 3 megabases (Mb).

The 3 Mb duplicated region contains 30 to 40 genes. For many of these genes, little is known about their function. A small percentage of affected individuals have a shorter duplication in the same region. Researchers are working to determine which duplicated genes may contribute to the developmental delay and other problems that sometimes affect people with this condition.

Learn more about the chromosome associated with 22q11.2 duplication

- chromosome 22

Inheritance

The inheritance of 22q11.2 duplication is considered autosomal dominant because the duplication affects one of the two copies of chromosome 22 in each cell. About 70 percent of affected individuals inherit the duplication from a parent. In other cases, the duplication is not inherited and instead occurs as a random event during the formation of reproductive cells (eggs and sperm) or in early fetal development. These affected people typically have no history of the disorder in their family, although they can pass the duplication to their children.

Other Names for This Condition

- Chromosome 22q11.2 duplication syndrome
- Chromosome 22q11.2 microduplication syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Chromosome 22q11.2 microduplication syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2675369/)

Genetic and Rare Diseases Information Center

- 22q11.2 duplication syndrome (https://rarediseases.info.nih.gov/diseases/10557/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- CHROMOSOME 22q11.2 DUPLICATION SYNDROME (https://omim.org/entry/608363)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%2822q11.2+duplication%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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