16p11.2 duplication

Description

16p11.2 duplication is a chromosomal change in which a small amount of genetic material within chromosome 16 is abnormally copied (duplicated). The duplication occurs near the middle of the chromosome at a location designated p11.2. This duplication can have a variety of effects. Common characteristics that occur in people with a 16p11.2 duplication include a low weight; a small head size (microcephaly); and developmental delay, especially in speech and language. Affected individuals also have an increased risk of behavioral problems. However, some people with the duplication have no identified physical or behavioral abnormalities.

Developmental delay and intellectual disability can occur in people with a 16p11.2 duplication. Approximately one-third of children with this condition have delays in developing physical skills such as sitting, crawling, or walking. The average IQ of affected individuals is about 26 points lower than that of their parents without the duplication. About 80 percent of people with a 16p11.2 duplication have problems related to speech or language. Both expressive language skills (vocabulary and the production of speech) and receptive language skills (the ability to understand speech) can be affected.

One of the most common behavioral problems associated with this chromosomal change is attention-deficit/hyperactivity disorder (ADHD). Autism spectrum disorder, which affects communication and social skills, is diagnosed in about one in five people with a 16p11.2 duplication. Affected individuals also have an increased risk of mental health problems, including schizophrenia, anxiety, and depression. Recurrent seizures are possible in this condition, although they do not occur in most affected individuals.

Other abnormalities that can occur with a 16p11.2 duplication include malformations of the kidneys and urinary tract. However, there is no particular pattern of physical abnormalities that characterizes 16p11.2 duplications; signs and symptoms related to the chromosomal change vary even among affected members of the same family.

Frequency

16p11.2 duplications have been estimated to occur in about 3 in 10,000 people. These changes are present in about 4 in 10,000 people who have mental health problems or difficulties with speech and language. Many people with the duplication are likely never diagnosed because there are many causes of these problems, and some people with

the duplication have no related health or developmental problems.

Causes

People with a 16p11.2 duplication have an extra copy of a segment of genetic material on the short (p) arm of chromosome 16 at a position known as p11.2. This duplication affects one of the two copies of chromosome 16 in each cell. The length of the duplicated segment is most often about 600,000 DNA building blocks (base pairs), also written as 600 kilobases (kb). The 600 kb region contains more than 25 genes, and little is known about the function of most of these genes. Researchers are working to determine how the extra genetic material contributes to the features of 16p11.2 duplication.

Learn more about the chromosome associated with 16p11.2 duplication

- chromosome 16

Inheritance

16p11.2 duplications have an autosomal dominant inheritance pattern, which means that a duplication in one copy of chromosome 16 in each cell is sufficient to cause the condition. Most affected individuals inherit the duplication from one affected parent; they may have similar characteristics of the condition as the parent, or they may be either more or less severely affected. However, in some cases 16p11.2 duplications are not inherited. Instead, they occur as random events during the formation of reproductive cells (eggs and sperm) or in early fetal development. People with a new duplication typically have no history of related signs or symptoms in their family, although their children can inherit the chromosomal change.

Other Names for This Condition

- 16p11.2 duplication syndrome
- 16p11.2 microduplication
- Autism, susceptibility to, 14B
- AUTS14B

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Chromosome 16p11.2 duplication syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3553407/)

Genetic and Rare Diseases Information Center

- Proximal 16p11.2 microduplication syndrome (https://rarediseases.info.nih.gov/diseases/12388/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%2216p11.2 duplication%22)

Catalog of Genes and Diseases from OMIM

- CHROMOSOME 16p11.2 DUPLICATION SYNDROME (https://omim.org/entry/614671)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%2816p11.2+duplication%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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