16p11.2 deletion syndrome

Description

16p11.2 deletion syndrome is a disorder caused by a deletion of a small piece of chromosome 16. The deletion occurs near the middle of the chromosome at a location designated p11.2.

People with 16p11.2 deletion syndrome usually have developmental delay and intellectual disability. Most also have at least some features of autism spectrum disorders. These disorders are characterized by impaired communication and socialization skills, as well as delayed development of speech and language. In 16p11.2 deletion syndrome, expressive language skills (vocabulary and the production of speech) are generally more severely affected than receptive language skills (the ability to understand speech). Some people with this disorder have recurrent seizures (epilepsy).

Some affected individuals have minor physical abnormalities such as low-set ears or partially webbed toes (partial syndactyly). People with this disorder are also at increased risk of obesity compared with the general population. However, there is no particular pattern of physical abnormalities that characterizes 16p11.2 deletion syndrome. Signs and symptoms of the disorder vary even among affected members of the same family. Some people with the deletion have no identified physical, intellectual, or behavioral abnormalities.

Frequency

Most people tested for the 16p11.2 deletion have come to medical attention as a result of developmental delay or autistic characteristics. Other individuals with the 16p11.2 deletion have no associated health or behavioral problems, and so the deletion may never be detected. For this reason, the prevalence of this deletion in the general population is difficult to determine but has been estimated at approximately 3 in 10,000.

Causes

People with 16p11.2 deletion syndrome are missing a sequence of about 600,000 DNA building blocks (base pairs), also written as 600 kilobases (kb), at position p11.2 on chromosome 16. This deletion affects one of the two copies of chromosome 16 in each cell. The 600 kb region contains more than 25 genes, and in many cases little is known about their function. Researchers are working to determine how the missing genes

contribute to the features of 16p11.2 deletion syndrome.

Learn more about the chromosome associated with 16p11.2 deletion syndrome

- chromosome 16

Inheritance

16p11.2 deletion syndrome is considered to have an autosomal dominant inheritance pattern because a deletion in one copy of chromosome 16 in each cell is sufficient to cause the condition. However, most cases of 16p11.2 deletion syndrome are not inherited. The deletion occurs most often as a random event during the formation of reproductive cells (eggs and sperm) or in early fetal development. Affected people typically have no history of the disorder in their family, although they can pass the condition to their children. Several examples of inherited 16p11.2 deletion have been reported. In inherited cases, other family members may be affected as well.

Other Names for This Condition

- Autism, susceptibility to, 14A
- AUTS14A

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Proximal 16p11.2 microdeletion syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3150154/)

Genetic and Rare Diseases Information Center

- Proximal 16p11.2 microdeletion syndrome (https://rarediseases.info.nih.gov/diseases/10740/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 deletion syndrome%22)

Catalog of Genes and Diseases from OMIM

- CHROMOSOME 16p11.2 DELETION SYNDROME, 593-KB (https://omim.org/entry/611913)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%2816p11.2+deletion%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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