15q13.3 Microdeletion

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15q13.3 microdeletion is a chromosomal change in which a small piece of chromosome 15 is deleted in each cell. The deletion occurs on the long (q) arm of the chromosome at a position designated q13.3. This chromosomal change increases the risk of intellectual disability, seizures, behavioral problems, and psychiatric disorders. However, some people with a 15q13.3 microdeletion do not appear to have any associated features.

Keywords: genetic conditions

1. Introduction

About half of all people with a 15q13.3 microdeletion have learning difficulties or intellectual disability, which is usually mild or moderate. Many of these individuals have delayed speech and language skills. 15q13.3 microdeletion also appears to be a major risk factor for recurrent seizures (epilepsy); about one-third of people with this chromosomal change have epilepsy.

15q13.3 microdeletion has also been associated with behavioral problems, including a short attention span, aggression, impulsive behavior, and hyperactivity. Some people with a 15q13.3 microdeletion have been diagnosed with developmental disorders that affect communication and social interaction (autism spectrum disorders). This chromosomal change may also be associated with an increased risk of psychiatric disorders, particularly schizophrenia. Other signs and symptoms of 15q13.3 microdeletion can include heart defects, minor abnormalities involving the hands and arms, and subtle differences in facial features.

Some people with a 15q13.3 microdeletion do not have any of the intellectual, behavioral, or physical features described above. In these individuals, the microdeletion is often detected when they undergo genetic testing because they have an affected relative. It is unknown why a 15q13.3 microdeletion causes cognitive and behavioral problems in some individuals but few or no health problems in others.

2. Frequency

15q13.3 microdeletion likely occurs in about 1 in 40,000 people in the general population. It appears to be more common in people with intellectual disability, epilepsy, schizophrenia, or autism spectrum disorders.

3. Causes

Most people with a 15q13.3 microdeletion are missing a sequence of about 2 million DNA building blocks (base pairs), also written as 2 megabases (Mb), at position q13.3 on chromosome 15. The exact size of the deleted region varies, but it typically contains at least six genes. This deletion usually affects one of the two copies of chromosome 15 in each cell.

The signs and symptoms that can result from a 15q13.3 microdeletion are probably related to the loss of one or more genes in this region. However, it is unclear which missing genes contribute to the specific features of the disorder. Because some people with a 15q13.3 microdeletion have no obvious signs or symptoms, researchers believe that other genetic or environmental factors may also be involved.

3.1. The chromosome associated with 15q13.3 microdeletion

- chromosome 15

4. Inheritance

15q13.3 microdeletion is inherited in an autosomal dominant pattern, which means one copy of the deleted region on chromosome 15 in each cell is sufficient to increase the risk of intellectual disability and other characteristic features.

In about 75 percent of cases, individuals with 15q13.3 microdeletion inherit the chromosomal change from a parent. In the remaining cases, 15q13.3 microdeletion occurs in people whose parents do not carry the chromosomal change. In these individuals, the deletion occurs most often as a random event during the formation of reproductive cells (eggs and sperm) or in early fetal development.

5. Other Names for This Condition

- 15q13.3 microdeletion syndrome
- chromosome 15q13.3 deletion syndrome

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