1q21.1 microdeletion

Description

1q21.1 microdeletion is a chromosomal change in which a small piece of chromosome 1 is deleted in each cell. The deletion occurs on the long (q) arm of the chromosome in a region designated q21.1. This chromosomal change increases the risk of delayed development, intellectual disability, physical abnormalities, and neurological and psychiatric problems. However, some people with a 1q21.1 microdeletion do not appear to have any associated features.

About 75 percent of all children with a 1q21.1 microdeletion have delayed development, particularly affecting the development of motor skills such as sitting, standing, and walking. The intellectual disability and learning problems associated with this genetic change are usually mild.

Distinctive facial features can also be associated with 1q21.1 microdeletions. The changes are usually subtle and can include a prominent forehead; a large, rounded nasal tip; a long space between the nose and upper lip (philtrum); and a high, arched roof of the mouth (palate). Other common signs and symptoms of 1q21.1 microdeletions include an unusually small head (microcephaly), short stature, and eye problems such as clouding of the lenses (cataracts). Less frequently, 1q21.1 microdeletions are associated with heart defects, abnormalities of the genitalia or urinary system, bone abnormalities (particularly in the hands and feet), and hearing loss.

Neurological problems that have been reported in people with a 1q21.1 microdeletion include seizures and weak muscle tone (hypotonia). Psychiatric or behavioral problems affect a small percentage of people with this genetic change. These include developmental conditions called autism spectrum disorders that affect communication and social interaction, attention-deficit/hyperactivity disorder (ADHD), and sleep disturbances. Studies suggest that deletions of genetic material from the 1q21.1 region may also be risk factors for schizophrenia.

Some people with a 1q21.1 microdeletion do not have any of the intellectual, physical, or psychiatric features described above. In these individuals, the microdeletion is often detected when they undergo genetic testing because they have a relative with the chromosomal change. It is unknown why 1q21.1 microdeletions cause cognitive and physical changes in some individuals but few or no health problems in others, even within the same family.

Frequency

1q21.1 microdeletion is a rare chromosomal change; only a few dozen individuals with this deletion have been reported in the medical literature.

Causes

Most people with a 1q21.1 microdeletion are missing a sequence of about 1.35 million DNA building blocks (base pairs), also written as 1.35 megabases (Mb), in the q21.1 region of chromosome 1. However, the exact size of the deleted region varies. This deletion affects one of the two copies of chromosome 1 in each cell.

The signs and symptoms that can result from a 1q21.1 microdeletion are probably related to the loss of several genes in this region. Researchers are working to determine which missing genes contribute to the specific features associated with the deletion. Because some people with a 1q21.1 microdeletion have no obvious related features, additional genetic or environmental factors are thought to be involved in the development of signs and symptoms.

Researchers sometimes refer to 1q21.1 microdeletion as the recurrent distal 1.35-Mb deletion to distinguish it from the genetic change that causes thrombocytopenia-absent radius syndrome (TAR syndrome). TAR syndrome results from the deletion of a different, smaller DNA segment in the chromosome 1q21.1 region near the area where the 1.35-Mb deletion occurs. The chromosomal change related to TAR syndrome is often called the 200-kb deletion.

Learn more about the chromosome associated with 1q21.1 microdeletion

- chromosome 1

Additional Information from NCBI Gene:

- ACP6
- BCL9
- CHD1L
- FMO5
- GJA5
- GJA8
- GPR89B
- HYDIN
- PRKAB2

Inheritance

1q21.1 microdeletion is inherited in an autosomal dominant pattern, which means that missing genetic material from one of the two copies of chromosome 1 in each cell is sufficient to increase the risk of delayed development, intellectual disability, and other signs and symptoms.

In at least half of cases, individuals with a 1q21.1 microdeletion inherit the chromosomal change from a parent. In general, parents who carry a 1q21.1 microdeletion have milder signs and symptoms than their children who inherit the deletion, even though the deletion is the same size. About one-quarter of these parents have no associated features.

A 1q21.1 microdeletion can also occur in people whose parents do not carry the chromosomal change. In this situation, the deletion occurs most often as a random event during the formation of reproductive cells (eggs or sperm) in a parent or in early embryonic development.

Other Names for This Condition

- 1q21.1 contiguous gene deletion
- 1q21.1 deletion
- Chromosome 1q21.1 deletion syndrome
- Chromosome 1q21.1 deletion syndrome, 1.35-Mb

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Chromosome 1q21.1 deletion syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2675897/)

Genetic and Rare Diseases Information Center

- 1q21.1 microdeletion syndrome (https://rarediseases.info.nih.gov/diseases/10813/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%221q21.1 microdeletion%

22)

Catalog of Genes and Diseases from OMIM

- CHROMOSOME 1q21.1 DELETION SYNDROME, 1.35-MB (https://omim.org/entry/612474)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%281q21.1%5BTIAB%5D%29+AND+%28%28microdeletion*%5BTIAB%5D%29+OR+%28deletion*%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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