17q12 duplication

Description

17q12 duplication is a chromosomal change in which a small piece of chromosome 17 is copied (duplicated) abnormally in each cell. The duplication occurs on the long (q) arm of the chromosome at a position designated q12.

Signs and symptoms related to 17q12 duplications vary significantly, even among members of the same family. Some individuals with the duplication have no apparent signs or symptoms, or the features are very mild. Other individuals can have intellectual disability, delayed development, and a wide range of physical abnormalities.

Intellectual and learning ability in people with 17q12 duplications ranges from normal to severely impaired. Many affected individuals have delayed development, particularly involving speech and language skills and gross motor skills such sitting, standing, and walking. Seizures are also common. Neurodevelopmental and psychiatric conditions that have been reported in people with 17q12 duplications include autism spectrum disorder (which affects social interaction and communication), schizophrenia, aggression, and self-injury. About half of affected individuals have an unusually small head (microcephaly).

Less commonly, 17q12 duplications have been associated with abnormalities of the eyes, heart, kidneys, and brain. Some individuals with this chromosomal change have subtle differences in facial features, although these are not consistent.

Frequency

17q12 duplications appear to be uncommon. Several dozen people with this chromosomal change have been described in the medical literature.

Causes

Most people with 17q12 duplications have an extra copy of about 1.4 million DNA building blocks (base pairs), also written as 1.4 megabases (Mb), at position q12 on chromosome 17. This duplication affects one of the two copies of chromosome 17 in each cell.

The duplicated segment is surrounded by short, repeated sequences of DNA that make the segment prone to rearrangement during cell division. The rearrangement can lead

to extra or missing copies of DNA at 17q12. (A missing copy of this segment causes a related chromosomal condition called 17q12 deletion syndrome.)

The segment of 17q12 that is most commonly duplicated includes at least 15 genes. It is unclear which of these genes, when present in more than one copy, contribute to intellectual disability, delayed development, and the other signs and symptoms described above. Because some people with a 17q12 duplication have no obvious intellectual or physical problems, researchers suspect that additional genetic factors may influence whether a person has signs and symptoms related to the chromosomal change.

Learn more about the chromosome associated with 17q12 duplication

- chromosome 17

Inheritance

17q12 duplications have an autosomal dominant pattern of inheritance, which means one copy of the duplication in each cell is sufficient to cause the signs and symptoms.

Most 17q12 duplications are inherited from a parent. In these cases, the parent most often has only mild signs and symptoms or no related features at all. Less commonly, 17q12 duplications represent a new (de novo) chromosomal change and occur in people with no history of the duplication in their family.

Other Names for This Condition

- 17q12 duplication syndrome
- 17q12 microduplication
- 17q12 microduplication syndrome
- 17q12 recurrent duplication
- Chromosome 17q12 duplication syndrome
- Recurrent duplication of 17q12

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Chromosome 17q12 duplication syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3281137/)

Genetic and Rare Diseases Information Center

- 17q12 microduplication syndrome (https://rarediseases.info.nih.gov/diseases/13296

/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%2217q12 duplication%22)

Catalog of Genes and Diseases from OMIM

- CHROMOSOME 17q12 DUPLICATION SYNDROME (https://omim.org/entry/614526)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%2817q12%5BTIAB%5D%29+AND+%28%28duplication%5BTIAB%5D%29+OR+%28microduplication%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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