3q29 microduplication syndrome

Description

3q29 microduplication syndrome (also known as 3q29 duplication syndrome) is a condition that results from the copying (duplication) of a small piece of chromosome 3 in each cell. The duplication occurs on the long (q) arm of the chromosome at a position designated q29.

The features associated with 3q29 microduplication syndrome vary widely. Some individuals with this chromosomal change have very mild or no related signs and symptoms, and the duplication is discovered because they undergo genetic testing only after a family member is diagnosed. Other people with a 3q29 microduplication have delayed development (particularly speech delay) and intellectual disability or learning difficulties. Although most affected individuals have no major birth defects, eye abnormalities, heart defects, and an unusually small head (microcephaly) can occur. 3q29 microduplication syndrome may increase the likelihood of being overweight or having obesity, although it is hard to determine whether these weight issues are caused by the duplication.

Frequency

3q29 microduplication syndrome appears to be very rare. Fewer than 30 affected individuals have been described in the medical literature.

Causes

Most people with 3q29 microduplication syndrome have an extra copy of about 1.6 million DNA building blocks (base pairs), also written as 1.6 megabases (Mb), at position q29 on chromosome 3. However, the duplication can vary in size. It affects one of the two copies of chromosome 3 in each cell.

The segment that gets duplicated is surrounded by short, repeated sequences of DNA that make it prone to rearrangement during cell division. The rearrangement can lead to missing or extra copies of DNA at 3q29. (A missing copy of this segment causes another condition called 3q29 microdeletion syndrome.)

The chromosome segment most commonly duplicated in people with 3q29 microduplication syndrome contains about 20 genes. Some of these genes are thought to be involved in brain and eye development. However, it is unknown which specific

genes, when abnormally copied, are related to the varied signs and symptoms of 3q29 microduplication syndrome. It is also unclear why some people with a duplication at 3q29 have no associated health problems. It is possible that genetic changes outside the 3q29 region can influence the features of this condition.

Learn more about the chromosome associated with 3q29 microduplication syndrome

- chromosome 3

Inheritance

This condition has an autosomal dominant pattern of inheritance, which means the duplication occurs on one copy of chromosome 3 in each cell.

In many cases, an affected person inherits the duplication from a parent. The parent may have no signs and symptoms related to the duplication, or the features may be mild. The remaining cases result from a new chromosomal change and occur in people with no history of the duplication in their family.

Other Names for This Condition

- 3q29 interstitial microduplication
- 3q29 microduplication
- Chromosome 3q29 duplication syndrome
- Microduplication 3q29 syndrome
- Trisomy 3q29

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Chromosome 3q29 microduplication syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2749873/)

Genetic and Rare Diseases Information Center

- 3q29 microduplication syndrome (https://rarediseases.info.nih.gov/diseases/10360/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- CHROMOSOME 3q29 DUPLICATION SYNDROME (https://omim.org/entry/611936)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%283q29%5BTI%5D%29+AND+%28*duplication*%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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