3q29 microdeletion syndrome

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

The features associated with 3q29 microdeletion syndrome vary widely. Some individuals with this chromosomal change have very mild or no related signs and symptoms, and the deletion is discovered through genetic testing only after a family member is diagnosed. However, most people with a 3q29 microdeletion have delayed development (particularly speech delay) and mild or moderate intellectual disability. They also have an increased risk of behavioral or psychiatric disorders, including autism spectrum disorder (which affects social interaction and communication), anxiety, bipolar disorder, and schizophrenia.

Infants with 3q29 microdeletion syndrome often have feeding difficulties and do not grow and gain weight at the expected rate (which is described as failure to thrive). Weak muscle tone (hypotonia), recurrent ear infections, an unusually small head (microcephaly), and yellowing of the skin and whites of the eyes (jaundice) can also occur. Some affected babies are born with a heart defect, most commonly an abnormal connection between two major arteries called patent ductus arteriosus (PDA).

Other possible features of 3q29 microdeletion syndrome include gastrointestinal disorders, such as a backflow of acidic stomach contents into the esophagus (gastroesophageal reflux), and abnormalities of the teeth. There may also be a subtle pattern of characteristic facial features, including a long, narrow face; a narrow space between the nose and upper lip (short philtrum); a high bridge of the nose; and large ears.

Frequency

3q29 microdeletion syndrome appears to be very rare. Based on a study from Iceland, the condition has an estimated incidence of 1 in 30,000 to 40,000 people in that population. About 75 affected individuals have been described in the medical literature.

Causes

Most people with 3q29 microdeletion syndrome are missing about 1.6 million DNA building blocks (base pairs), also written as 1.6 megabases (Mb), at position q29 on chromosome 3. This deletion affects one of the two copies of chromosome 3 in each cell.

The segment that gets deleted 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. (An extra copy of this segment causes another condition called 3q29 microduplication syndrome.)

The chromosome segment most commonly deleted in people with 3q29 microdeletion syndrome contains about 20 genes. Some of these genes are thought to be involved in brain development. However, it is unknown which specific genes, when deleted, are related to the signs and symptoms of 3q29 microdeletion syndrome. It is also unclear why some people with a deletion at 3q29 have no associated health problems. It is possible that genetic changes outside the 3q29 region can influence the features of this condition.

**Inheritance Pattern**

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

Most cases of 3q29 microdeletion syndrome result from a new (de novo) chromosomal change and occur in people with no history of the deletion in their family. Less commonly, an affected person inherits the deletion from a parent. The parent may have no signs and symptoms related to the deletion, or the features may be mild.

**Other Names for This Condition**

- 3q subtelomere deletion syndrome
- 3q29 deletion syndrome
- 3q29 recurrent deletion
- chromosome 3q29 deletion syndrome
- microdeletion 3q29 syndrome
- monosomy 3q29

**Diagnosis & Management**

**Genetic Testing Information**

- What is genetic testing? /primer/testing/genetictesting
- Genetic Testing Registry: 3q29 microdeletion syndrome https://www.ncbi.nlm.nih.gov/gtr/conditions/C2674949/

**Research Studies from ClinicalTrials.gov**

- ClinicalTrials.gov https://clinicaltrials.gov/ct2/results?cond=%223q29+microdeletion+syndrome%22+OR+%223q29+deletion+syndrome%22
Other Diagnosis and Management Resources

• Emory University: 3q29 Project
  http://genome.emory.edu/3q29/

• GeneReview: 3q29 Recurrent Deletion
  https://www.ncbi.nlm.nih.gov/books/NBK385289

Additional Information & Resources

Health Information from MedlinePlus

• Encyclopedia: Gastroesophageal Reflux Disease
  https://medlineplus.gov/ency/article/000265.htm

• Encyclopedia: Patent Ductus Arteriosus
  https://medlineplus.gov/ency/article/001560.htm

• Health Topic: Autism Spectrum Disorder
  https://medlineplus.gov/autismspectrumdisorder.html

• Health Topic: Bipolar Disorder
  https://medlineplus.gov/bipolardisorder.html

• Health Topic: Developmental Disabilities
  https://medlineplus.gov/developmentaldisabilities.html

• Health Topic: Schizophrenia
  https://medlineplus.gov/schizophrenia.html

Genetic and Rare Diseases Information Center

• 3q29 microdeletion syndrome
  https://rarediseases.info.nih.gov/diseases/11974/3q29-microdeletion-syndrome

Additional NIH Resources

• Eunice Kennedy Shriver National Institute of Child Health and Human Development: Intellectual and Developmental Disabilities
  https://www.nichd.nih.gov/health/topics/idds

• National Human Genome Research Institute: Chromosome Abnormalities
  https://www.genome.gov/about-genomics/fact-sheets/Chromosome-Abnormalities-Fact-Sheet

• National Institute of Mental Health (NIMH): Anxiety Disorders
  https://www.nimh.nih.gov/health/topics/anxiety-disorders/index.shtml

• National Institute of Mental Health (NIMH): Bipolar Disorder
  https://www.nimh.nih.gov/health/topics/bipolar-disorder/index.shtml

• National Institute of Mental Health (NIMH): Schizophrenia
  https://www.nimh.nih.gov/health/topics/schizophrenia/index.shtml
Educational Resources

- MalaCards: chromosome 3q29 deletion syndrome
  https://www.malacards.org/card/chromosome_3q29_deletion_syndrome
- MentalHealth.gov
  https://www.mentalhealth.gov/what-to-look-for
- Orphanet: Orphanet: 3q29 microdeletion syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=65286
- Unique: 3q29 Deletions and Microdeletions
  https://www.rarechromo.org/media/information/Chromosome%20%203/3q29%20deletions%20and%20microdeletions%20FTNW.pdf

Patient Support and Advocacy Resources

- 3q29 Deletion and 3q29 Duplication Patient Registry
  https://3q29deletion.patientcrossroads.org/
- American Association on Intellectual and Developmental Disabilities
  https://www.aaidd.org/
- Chromosome Disorder Outreach
  https://chromodisorder.org/
- National Alliance on Mental Illness
  https://www.nami.org/Home
- Unique: The Rare Chromosome Disorder Support Group (UK)
  https://www.rarechromo.org/

Clinical Information from GeneReviews

- 3q29 Recurrent Deletion
  https://www.ncbi.nlm.nih.gov/books/NBK385289

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- CHROMOSOME 3q29 DELETION SYNDROME
  http://omim.org/entry/609425

Medical Genetics Database from MedGen

- 3q29 microdeletion syndrome
  https://www.ncbi.nlm.nih.gov/medgen/393265
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