15q24 Microdeletion

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15q24 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 q24.

Keywords: genetic conditions

1. Introduction

15q24 microdeletion is associated with mild to moderate intellectual disability and delayed speech development. Other common signs and symptoms include short stature, weak muscle tone (hypotonia), and skeletal abnormalities including loose (lax) joints. Affected males may have genital abnormalities, which can include an unusually small penis (micropenis) and the opening of the urethra on the underside of the penis (hypospadias). Affected individuals also have distinctive facial features such as a high front hairline, broad eyebrows, widely set eyes (hypertelorism), outside corners of the eyes that point downward (downslanting palpebral fissures), a broad nasal bridge, a full lower lip, and a long, smooth space between the upper lip and nose (philtrum).

2. Frequency

This condition is very rare; only a few dozen affected individuals have been identified.

3. Causes

People with a 15q24 microdeletion are missing between 1.7 million and 6.1 million DNA building blocks (base pairs), also written as 1.7-6.1 megabases (Mb), at position q24 on chromosome 15. The exact size of the deletion varies, but all individuals are missing the same 1.2 Mb region. This region contains several genes that are thought to be important for normal development.

The signs and symptoms that result from a 15q24 microdeletion are probably related to the loss of one or more genes in the deleted region. However, it is unclear which missing genes contribute to the specific features of the disorder.

3.1. The chromosome associated with 15q24 microdeletion

- chromosome 15

4. Inheritance

The identified cases of 15q24 microdeletion have occurred in people with no history of the condition in their family. The chromosomal change likely occurs as a random event during the formation of reproductive cells (eggs or sperm) or in early fetal development.

5. Other Names for This Condition

- 15q24 deletion
- 15q24 microdeletion syndrome
- interstitial deletion of chromosome 15q24

References

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