2q37 Deletion Syndrome

Subjects: Genetics & Heredity
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2q37 deletion syndrome is a condition that can affect many parts of the body. Most babies with 2q37 deletion syndrome are born with weak muscle tone (hypotonia), which usually improves with age. Other neurological abnormalities that are common in affected individuals include mild to severe intellectual disability; delayed development of motor skills, such as sitting and walking; and behavioral problems. About 25 percent of people with this condition have autism spectrum disorder, a developmental condition that affects communication and social interaction.

Keywords: genetic conditions

1. Introduction

Unusual physical features are also common in people with 2q37 deletion syndrome. About half of affected individuals have unusually short fingers and toes (brachydactyly), often with abnormally short fourth toes that may overlap the other toes. Additional features of this condition may include short stature, obesity, or sparse hair. Many people with 2q37 deletion syndrome have characteristic facial features that can include a prominent forehead, a low frontal hairline, thin eyelids, skin folds covering the inner corner of the eyes (epicanthal folds), outside corners of the eyes that point upward (upslanting palpebral fissures), a small nose, a small mouth with thin lips, a smooth space between the upper lip and nose (smooth philtrum), prominent cheekbones, a large chin, and minor ear abnormalities.

Other features of 2q37 deletion syndrome can include seizures and an inflammatory skin disorder called eczema. Some affected individuals have malformations of the brain, heart, gastrointestinal system, kidneys, or genitalia. A few people with 2q37 deletion syndrome develop a rare form of kidney cancer called Wilms tumor.

2. Frequency

2q37 deletion syndrome appears to be a rare condition, although its exact prevalence is unknown. At least 115 cases have been reported worldwide.

3. Causes

2q37 deletion syndrome is caused by deletions of genetic material from a specific region in the long (q) arm of chromosome 2. The deletions occur near the end of the chromosome at a location designated 2q37. The size of the deletion varies among affected individuals, with most affected people missing 2 million to 9 million DNA building blocks (also written as 2 Mb to 9 Mb).

Researchers are working to identify all of the genes whose loss contributes to the features of 2q37 deletion syndrome. Many of these genes have not been well characterized. However, genes in this region appear to be critical for the normal development of many parts of the body.

Researchers have determined that loss of a particular gene on chromosome 2, called *HDAC4*, is likely to account for many of the syndrome's characteristic signs (such as intellectual disability and skeletal abnormalities). While the deleted segment in 2q37 deletion syndrome varies in size, it always contains the *HDAC4* gene. Additionally, a few people with mutations in only the *HDAC4* gene have many of the features of 2q37 deletion syndrome. It is unclear what role the other genes on 2q37 play in this disorder.

3.1. The gene and chromosome associated with 2q37 deletion syndrome

- HDAC4
- chromosome 2

4. Inheritance

Most cases of 2q37 deletion syndrome are not inherited. They result from a chromosomal deletion that occurs as a random event during the formation of reproductive cells (eggs or sperm) or in early fetal development. Affected people typically have no history of the disorder in their family.

Rarely, an affected individual inherits a copy of chromosome 2 with a deleted segment from an affected parent. In these cases, the parent is usually less severely affected than the child, for reasons that are unknown. When an affected child inherits a chromosomal deletion from a parent, it is inherited in an autosomal dominant pattern, which means one copy of the altered chromosome in each cell is sufficient to cause the disorder.

5. Other Names for This Condition

- 2q37 microdeletion syndrome
- Albright hereditary osteodystrophy-like syndrome
- brachydactyly-mental retardation syndrome
- chromosome 2q37 deletion syndrome (disorder)
- deletion 2q37
- monosomy 2q37

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