1p36 Deletion Syndrome

Subjects: Genetics & Heredity
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1p36 deletion syndrome is a disorder that typically causes severe intellectual disability. Most affected individuals do not speak, or speak only a few words. They may have temper tantrums, bite themselves, or exhibit other behavior problems. Most have structural abnormalities of the brain, and seizures occur in more than half of individuals with this disorder. Affected individuals usually have weak muscle tone (hypotonia) and swallowing difficulties (dysphagia).

Keywords: genetic conditions

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1. Introduction

People with 1p36 deletion syndrome have a small head that is also unusually short and wide in proportion to its size (microbrachycephaly). Affected individuals also have distinctive facial features including deep-set eyes with straight eyebrows; a sunken appearance of the middle of the face (midface hypoplasia); a broad, flat nose; a long area between the nose and mouth (philtrum); a pointed chin; and ears that are low-set, rotated backwards, and abnormally shaped.

People with 1p36 deletion syndrome may have vision or hearing problems. Some have abnormalities of the skeleton, heart, gastrointestinal system, kidneys, or genitalia.

2. Frequency

1p36 deletion syndrome is believed to affect between 1 in 5,000 and 1 in 10,000 newborns. However, this may be an underestimate because some affected individuals are likely never diagnosed.

3. Causes

1p36 deletion syndrome is caused by a deletion of genetic material from a specific region in the short (p) arm of chromosome 1. The signs and symptoms of 1p36 deletion syndrome are probably related to the loss of multiple genes in this region. The size of the deletion varies among affected individuals.

3.1. The chromosome associated with 1p36 deletion syndrome

- chromosome 1

4. Inheritance

Most cases of 1p36 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.

About 20 percent of people with 1p36 deletion syndrome inherit the chromosome with a deleted segment from an unaffected parent. In these cases, the parent carries a chromosomal rearrangement called a balanced translocation, in which no genetic material is gained or lost. Balanced translocations usually do not cause any health problems; however, they can become unbalanced as they are passed to the next generation. Children who inherit an unbalanced translocation can have a chromosomal rearrangement with extra or missing genetic material. Individuals with 1p36 deletion syndrome who inherit an unbalanced translocation are missing genetic material from the short arm of chromosome 1, which results in birth defects and other health problems characteristic of this disorder.

5. Other Names for This Condition

- chromosome 1p36 deletion syndrome

- distal monosomy 1p36
- monosomy 1p36 syndrome

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