1p36 deletion syndrome

Description

1p36 deletion syndrome is a disorder that typically causes intellectual disabilities. 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 difficulty swallowing (dysphagia).

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 often have unusually short fingers and toes (brachydactyly), permanently bent fingers and toes (camptodactyly), and short feet. They may also have vision or hearing problems. Some affected individuals have abnormalities of the skeleton, heart, gastrointestinal system, kidneys, or genitalia.

Life expectancy varies in people with 1p36 deletion syndrome, but affected individuals can survive into early adulthood.

Frequency

1p36 deletion syndrome is estimated to affect 1 in 5,000 newborns each year in the United States.

Causes

1p36 deletion syndrome is caused by the deletion of a piece of genetic material from a specific region in the short (p) arm of chromosome 1. In about 50 percent of people with this condition, the deleted region includes the tip of the p arm of chromosome 1. Around 29 percent of affected individuals have deletions that include a section near the end of the chromosome. The remaining 21 percent have complex rearrangements of genetic material that involve this region.

The size of the deletion varies among affected individuals. .cf0{font-style:italic;font-family:Segoe UI;font-size:9pt;}The signs and symptoms of 1p36 deletion syndrome are likely caused by the loss of multiple genes that are involved in the development of systems such as the brain, heart, and skeleton.

Learn more about the genes and chromosome associated with 1p36 deletion syndrome

- MTOR
- SKI
- chromosome 1

Additional Information from NCBI Gene:

- CDC42
- ECE1
- GABRD
- GNB1
- KCNAB2
- PRDM16
- RERE

Inheritance

Most cases of 1p36 deletion syndrome are not inherited. They are the result of 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. No genetic material is gained or lost in a balanced translocation, so these chromosomal changes usually do not cause any health problems. However, translocations 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 p arm of chromosome 1, which results in the health problems that are characteristic of this disorder.

Other Names for This Condition

- Chromosome 1p36 deletion syndrome
- Distal monosomy 1p36
- Monosomy 1p36 syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Chromosome 1p36 deletion syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1842870/)

Genetic and Rare Diseases Information Center

- 1p36 deletion syndrome (https://rarediseases.info.nih.gov/diseases/6082/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%221p36 deletion syndrome%22)

Catalog of Genes and Diseases from OMIM

- CHROMOSOME 1p36 DELETION SYNDROME, DISTAL (https://omim.org/entry/607872)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=((1p36+deletion+syndrome%5BTIAB%5D)+OR+(monosomy+1p36+syndrome%5BTIAB%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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