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My46 Trait Profile

1p36 deletion syndrome

Other Names: Monosomy 1p36

1p36 deletion syndrome results from a loss of genetic material on one of the short arms of chromosome 1. Deletions can vary in size, but typically result in a spectrum of physical, developmental, and behavioral characteristics.

Characteristics of 1p36 deletion syndrome

Individuals with 1p36 deletion syndrome often have characteristic facial features multiple congenital anomalies (i.e., birth defects), as well as developmental problems.

Facial features commonly seen in individuals with 1p36 deletion syndrome include straight eyebrows, deep-set eyes, midface hypoplasia (underdevelopment of jaw, cheekbones, and eye socket), broad nasal root/bridge, long philtrum (part between the upper lip and nose), pointed chin, low-set ears, large anterior fontanelle (soft spot in the skull of newborns and young babies), epicanthal folds (extra skin over the inside corner of the eye), microcephaly (small head), and brachycephaly (broad head). Less common physical features may include synophrys (thick, joined eyebrows), hypertelorism (extra space between the eyes), low hairline, small, upslanting, or downslanting palpebral fissures (distance between the inner and outer corners of one's eye), microstomia (small mouth), cleft lip/palate and a high palate.

Seizures are common among individuals with 1p36 deletion syndrome, though the age of onset and type of seizures vary. Congenital heart defects are present in approximately 50% of affected individuals; some people may develop cardiomyopathy as well. Vision problems may include strabismus (cross-eyed), near-sightedness, or far-sightedness; hearing loss (both conductive and sensorineural) is also common. Hypotonia (low muscle tone) is also common, which may contribute to developmental delay.

Some degree of global developmental delay is almost universal among people with 1p36 deletion syndrome; typically, speech development is most severely affected and individuals may not be able to verbally communicate until late childhood. Intellectual disability is usually present, though the degree ranges in severity; behavioral concerns including hyperphagia (over-eating without feeling full), autism-like behavior, and self-injurious behavior may also be present in approximately 30% of affected individuals.

Diagnosis/Testing

1p36 deletion syndrome is caused by a deletion (missing piece) of genetic material on one of the two copies of chromosome 1 in each cell. A microarray (also known as an oligoarray, SNP array, or arrayCGH) is a blood test that can simultaneously evaluate the cells for small pieces of genetic material that may be missing or extra on each chromosome (the packages of genetic material). A blood test known as FISH (fluorescence in situ hybridization) involves attaching fluorescent probes to the specific area of interest and is frequently used to confirm a deletion found through a microarray, and for testing family members of people with 1p36 deletion syndrome. While the deletion size may vary, most often it is about 4-5 Mb in size. It is not believed that the size of deletion correlates with how many characteristics are seen in someone with 1p36 deletion syndrome.

Management/Surveillance

Management of individuals with 1p36 deletion syndrome often includes care from multiple specialists due to the various medical and developmental concerns that may arise over time. Evaluation by a geneticist would be recommended. Intensive speech, physical, and occupational therapies are recommended to improve developmental outcome. Audiologic, ophthalmologic, and cardiac evaluations are recommended to screen for any problems with the individual's hearing, vision, and heart. In addition, the following specialties may also be involved in the care of someone with 1p36 deletion syndrome: neurology, gastroenterology and feeding, and orthopedics.

Mode of inheritance

1p36 deletion syndrome is inherited in an autosomal dominant pattern. This means inheriting one 1p36 deletion is enough for an individual to be affected and show signs of 1p36 deletion syndrome. Most of the time (approximately 95%), the deletion occurs brand new (de novo) in the child (de novo). However, it is also possible that a parent of a child with 1p36 deletion syndrome may carry a chromosome rearrangement that may predispose the child and future children to have 1p36 deletion syndrome.

Risk to family members

The risk to family members depends on whether or not the parent of the individual with 1p36 deletion syndrome carries a chromosome rearrangement that increases the chance of having a child with the deletion syndrome. If parental testing is normal, the risk of having another child with 1p36 deletion syndrome is very low.

Special considerations

None

Resources

1p36 Deletion Support and Awareness
http://www.1p36dsa.org/
Unique: Understanding chromosome disorders
http://www.rarechromo.org/information/Chromosome%20%201/1p36%20deletions%20FTNP.pdf
Genetics Home Reference: 1p36 deletion syndrome
http://ghr.nlm.nih.gov/condition/1p36-deletion-syndrome

References

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