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My46 Trait Profile

15q24 deletion syndrome

15q24 deletion syndrome results from a loss of genetic material on one of the long arms of chromosome 15. This deletion results in the loss of several genes.

Characteristics of 15q24 deletion syndrome

15q24 deletion syndrome is characterized by growth delay, feeding difficulties, and distinct facial features including a long face with high frontal hairline, wide-spaced eyes, sparse and broad eyebrows, and ear abnormalities. Skeletal and digital deformities have been described in the majority of individuals and may include loose joints, curvature of the spine (scoliosis), thumb and toe anomalies, and short digits (brachydactyly). Genital abnormalities such as hypospadias, small penis, and undescended testes have been reported in a majority of males with 15q24 deletion syndrome, however, genital abnormalities do not seem to occur with the same frequency in females with this deletion. Low muscle tone (hypotonia), which is found in over half of the reported cases, may contribute to the developmental delay which has been observed in nearly all of the reported individuals with 15q24 deletion syndrome. Varying degrees of intellectual disability, typically in the range of mild to moderate in severity, have been reported in nearly all individuals, while behavior challenges such as aggression, hyperactivity, autistic behaviors, and attention deficit disorder have been reported in over one-third of individuals.

Diagnosis/Testing

This condition is caused by a deletion (missing piece) of genetic material on one of the two copies of chromosome 15 in each cell. A microarray (also known as an oligoarray, SNP array or arrayCGH) is a blood test which 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 for testing family members of affected individuals.

15q24 deletion syndrome is caused by a small deletion on chromosome 15 that ranges in size from 1.7-6.1 Mb. This region contains several genes that may contribute to the features observed in individuals with this deletion. It is thought that the range of deletion sizes that can cause 15q24 deletion syndrome may partially explain the wide variability that can be seen within this condition.

Management/Surveillance

Management of individuals with 15q24 deletion syndrome often includes care from multiple specialists because of the various medical and developmental concerns that may arise over time. The following evaluations are recommended at the time of diagnosis: genetics evaluation, developmental assessment, hearing evaluation, eye examination, and heart ultrasound (echocardiogram) to evaluate for structural heart problems. Other specialists that may be involved in the care of individuals with 15q24 deletion include: allergy and immunology, endocrinology, gastroenterology and feeding, neurology, orthopedics, and urology.

Mode of inheritance

15q24 deletion syndrome is inherited in an autosomal dominant pattern. This means inheriting one 15q24 deletion is

enough for an individual to be affected and show signs of 15q24 deletion syndrome. The deletion can be inherited from an affected parent, however it most often occurs brand new (de novo) in an affected child. It is also possible that a parent of a child with this deletion syndrome could carry a chromosome rearrangement that may predispose to the deletion in the child.

Risk to family members

The risk to family members depends on whether or not the parent of the individual with 15q24 deletion carries a chromosome rearrangement that increases the chance of having a child with the deletion. If parental testing is normal, then the risk of having another child with 15q24 deletion syndrome is very low.

Special considerations

None

Resources

Genetics Home Reference: 15q24 deletion syndrome
http://ghr.nlm.nih.gov/condition/15q24-microdeletion
Unique: 15q24 deletion syndrome
http://www.rarechromo.org/information/Chromosome%2015/15q24%20microdeletion%20syndrome%20FTNP.pdf

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