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My46 Trait Profile

16p11.2 microdeletion

16p11.2 microdeletion results from a loss of genetic material on one of the short arms of chromosome 16. This deletion results in the loss of several genes.

Characteristics of 16p11.2 microdeletion

16p11.2 microdeletion is quite variable depending on the size of the deletion as well as other genetic and environmental factors. However, most individuals with 16p11.2 microdeletion have developmental delays (most notably with communication and cognitive skills) and intellectual disability. Most, but not all, also have some features of autism spectrum disorder. Additional features include attention deficit hyperactivity disorder, obesity, seizures, and macrocephaly (larger than expected head size). Although the majority of individuals have not been reported to have specific birth defects, there have been individuals with mild heart defects as well as other minor physical differences.

Diagnosis/Testing

This condition is caused by a deletion (missing piece) of genetic material on one of the two copies of chromosome 16 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 (flourescence in situ hybridization) involves attaching fluorescent probes to the specific area of interest and is frequently used for confirmation or testing family members of affected individuals.

Management/Surveillance

Management of 16p11.2 microdeletion often involves regular development assessments and educational interventions. A heart evaluation is often suggested at the time of diagnosis since some individuals may have minor heart defects.. Other monitoring may be necessary given each child’s unique symptoms.

Mode of inheritance

16p11.2 microdeletion is inherited in an autosomal dominant pattern. This means inheriting one 16p11.2 deletion is usually enough for an individual to be affected and show signs of 16p11.2 microdeletion. The deletion can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not the individual with 16p11.2 microdeletion has a parent with the deletion. If a parent also has the deletion, the risk for that parent to have another child with the deletion is 50% with each pregnancy. If a parent does not have the deletion, the risk of other siblings being affected is less than 1%.

Special considerations

None

Resources

Genetics Home Reference: 16p11.2 deletion syndrome
http://ghr.nlm.nih.gov/condition/16p112-deletion-syndrome
Simons VIP Connect Registry
http://www.simonsvipconnect.org/forms/GeneticsFactSheet16Del.pdf
Unique: Understanding Chromosome Disorders
http://www.rarechromo.org/forum/LeafletConfirm.asp?ch=Chromosome%2016&fn=16p11.2%20microdeletions%20FTNI

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