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My46 Trait Profile

16p11.2 microduplication

16p11.2 microduplication results from a gain of genetic material on one of the short arms of chromosome 16. This duplication results in the gain of several genes.

Characteristics of 16p11.2 microduplication

16p11.2 microduplication is quite variable depending on the size of the duplication as well as other genetic and environmental factors. Individuals with 16p11.2 microduplication have an increased risk for developmental delays (most notably with communication and cognitive skills), neuropsychological concerns such as attention deficit hyperactivity disorder and autism, failure to thrive, being underweight, and microcephaly (smaller than expected head size). Although the majority of individuals have not been reported to have specific birth defects, there have been a few individuals with various birth defects including kidney, brain, and diaphragm abnormalities.

Diagnosis/Testing

This condition is caused by a duplication (extra 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 microduplication often involves regular development assessments and educational interventions. Other monitoring may be necessary given each child’s unique symptoms.

Mode of inheritance

16p11.2 microduplication is inherited in an autosomal dominant pattern. This means inheriting one 16p11.2 duplication is usually enough for an individual to be affected and show signs of 16p11.2 microduplication. The duplication 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 microduplication has a parent with the duplication. If a parent also has the duplication, the risk for that parent to have another child with the duplication is 50% with each pregnancy. If a parent does not have the duplication, the risk of other siblings being affected is less than 1%.

Special considerations

None

Resources

Simons VIP Connect Registry

http://www.simonsvipconnect.org/forms/GeneticsFactSheet16Dup.pdf
Unique: Understanding Chromosome Disorders
http://www.rarechromo.org/information/Chromosome%2016/16p11.2%20microduplications%20FTNP.pdf

References

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