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My46 Trait Profile

1q21.1 microduplication

1q21.1 microduplication results from a gain of genetic material on one of the long arms of chromosome 1. This duplication results in the gain of several genes.

Characteristics of 1q21.1 microduplication

1q21.1 microduplication is quite variable depending on the size of the duplication as well as other genetic and environmental factors. Some individuals have no obvious birth defects with normal developmental progress and normal intelligence. However, individuals with 1q21.1 microduplication do have an increased risk for developmental delays (typically mild), autistic features, larger than average head size, and in some cases heart defects or psychiatric concerns such as schizophrenia.

Diagnosis/Testing

This condition is caused by a duplication (extra 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 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.

Management/Surveillance

Development assessments, as well as evaluations of the heart are suggested at the time of diagnosis if not already performed. Other monitoring may be necessary given each child's unique symptoms. Other monitoring may be necessary given each child's unique symptoms.

Mode of inheritance

1q21.1 microduplication is inherited in an autosomal dominant pattern. This means inheriting one 1q21.1 duplication is usually enough for an individual to be affected and show signs of 1q21.1 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 1q21.1 microduplication has a parent with the duplication. If a parent also has the duplication, the risk for that parent to have another with the duplication is 50% with each pregnancy. If a parent does not have the duplication, the risk of other siblings being affected is very low.

Special considerations

None

Resources

Simons VIP Connect Registry
http://www.simonsvipconnect.org/forms/FactSheet1qDuplication.pdf

Unique: Understanding Chromosome Disorders
http://www.rarechromo.org/information/Chromosome%20%201/1q21.1%20microduplications%20FTNW.pdf

References

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Created: 03/2013 **Created by:** Megan Tucker, MS, LGC

Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC