Published on *My46* (https://www.my46.org) 10/16/2021

My46 Trait Profile

Koolen-De Vries syndrome

Other Names: 17q21.31 microdeletion

Koolen-De Vries syndrome is a rare genetic condition with characteristic facial features, birth defects, and developmental delays. It is caused by mutations in the KANSL1 gene which makes a protein that is part of the KAT8 regulatory NSL complex.

Characteristics of Koolen-De Vries syndrome

Koolen-De Vries syndrome is a genetic condition with characteristic facial features such as a high or broad forehead, long face, tubular shaped nose, large ears, and multiple congenital anomalies such as heart, kidney, and gastrointestinal abnormalities, and hypotonia (low muscle tone). Infants with Koolen-De Vries syndrome also tend to have poor weight gain. Some affected individuals have epilepsy. Varying degrees of developmental delay and/or intellectual disability are also commonly seen in individuals with this syndrome.

Diagnosis/Testing

Koolen-De Vries syndrome is caused by changes or mutations in the KANSL1 gene. This gene makes the KAT8 regulatory NSL complex subunit 1 protein. This protein is involved in controlling the activity of genes that are important for normal development and function. Most individuals with Koolen-De Vries syndrome have a deletion (missing piece) of one of the two copies of the KANSL1 genes. 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 confirmation or testing family members of affected individuals.

Management/Surveillance

Management of the condition often involves regular developmental assessments and educational interventions. Other monitoring is typically guided by each individual's unique symptoms such as an assessment of cardiac or kidney problems in addition to evaluating for seizure activity.

Mode of inheritance

Koolen-De Vries syndrome is inherited in an autosomal dominant pattern. This means inheriting one KANSL1 mutation is enough for an individual to be affected and show signs of the condition. The deletion can be inherited from an affected parent, however all cases to date have occurred 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 Koolen-De Vries syndrome has a parent with the condition. To date all individuals with Koolen-De Vries syndrome have the condition as a result of a de novo mutation. If a parent does not have Koolen-De Vries syndrome, the risk of other siblings being affected is very low.

Special considerations

None

Resources

Genetics Home Reference: Koolen-de Vries syndrome
http://ghr.nlm.nih.gov/condition/koolen-de-vries-syndrome
Unique: Understanding Chromosome Disorders
http://www.rarechromo.org/information/Chromosome%2017/Koolen-De%20Vries%20Syndrome%20FTNP.pdf
17q21.31 research project
http://17q21.com/

References

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Created: 03/2013 **Created by:** Megan Tucker, MS, LGC
Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC