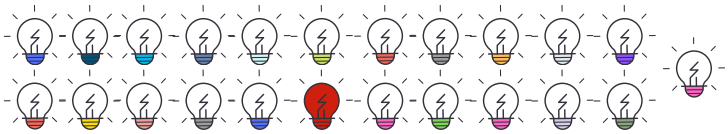
What are 17q12 CNVs?

The 17q12 Foundation represents two separate syndromes:

17q12 deletion syndrome



is caused by a missing piece of chromosome 17 (deletion) that is present from the moment the child is conceived

17q12 duplication syndrome

is caused by an extra piece of chromosome 17 (microduplication) that is present from the moment the child is conceived

1 in 14500 people

in the general population have this deletion syndrome. It's more common in populations with developmental disorders developmental delay, intellectual disability autism spectrum disorder) and schizophrenia

The deletion is most often a brand new (de novo), sporadic event in the person that is diagnosed

of the time a person with the deletion will have inherited it from a parent

chance that each of that parent's children will also have the deletion

chances that a sibling has if it was de novo (both parents tested negative)

1 in 2500 individuals

in the general population have this duplication syndrome. It's more common in populations with developmental disorders (developmental delay, intellectual disability autism spectrum disorder) and schizophrenia

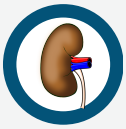
The duplication is most often inherited from a parent. Oftentimes, the duplication is only identified in parents after the child is diagnosed and could have similar, milder or apparently no features

chances that each of that parent's children will also have the duplication

of people with the duplication will have a brand new (de novo) duplication that wasn't inherited from either parent

chances that a sibling has the duplication if it was de novo (both parents tested negative)

A syndrome is defined as a recognizable group of signs and symptoms that consistently occur together.



Kidney and urinary tract abnormalities

Macrocephaly (Large head size)

Maturity onset diabetes of the young type 5 (MODY-5)

Neurodevelopmental/Psychiatric: developmental delay, autism spectrum disorder, learning disabi-lity, intellectual disability, anxiety, bipolar disorder, schizophrenia

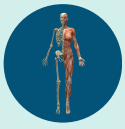
It's important to remember that no two people with the deletion will have the same combination and/or severity of symptoms, even people within the same family

The most common features of the duplication are related to neurodevelopment.

Skeletal differences

Microcephaly (small head size)

Seizures in up to 75% of individuals



Hypotonia (low muscle tone)

Neurodevelopmental/Psychiatric: intellectual abilities ranging from typical to severe disability, speech delay, motor delay, behavioral concerns (aggression, compulsive disorders), autism spectrum disorder

It is important to remember that no two people with the duplication will have the same combination and/or severity of symptoms, even people within the same family

For More Information
visit www.chromo17q12.org/

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