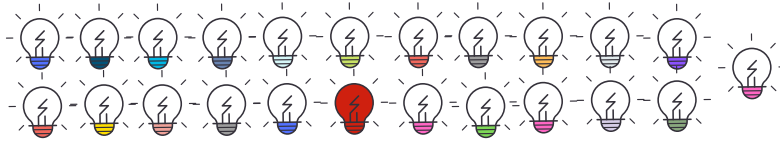


What are 17q12 CNVs?

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

 **in 14500 people**

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

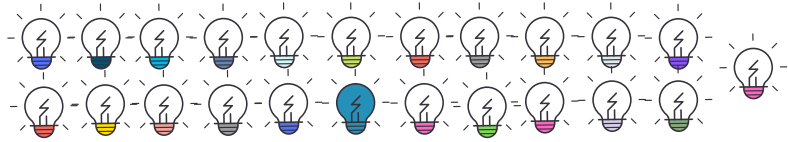


chance that each of that parent's children will also have the deletion



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

17q12 duplication syndrome



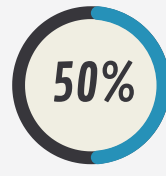
is caused by an extra piece of chromosome 17 (microduplication) that is present from the moment the child is conceived

 **in 2500 individuals**

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



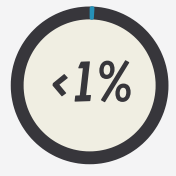
The duplication is most often inherited from a parent. Oftentimes, the duplication is only identified in parents after the child is diagnosed and could have similar, milder or apparently no features



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

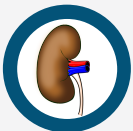


of people with the duplication will have a brand new (*de novo*) duplication that was not inherited from either parent



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 disability, intellectual disability, anxiety, bipolar disorder, schizophrenia

It is 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/

INFOGRAM MADE BY PRISMA RESEARCH