17q12 FOUNDATION

NEWSLETTER

NO. 1 • 2019

17q12

5 WAYS TO COOK YOUR EGGS

Never make something boring ever again | 18

IT'S ABOUT THE INGREDIENTS

Chef Leah gives tips on how to pick the best ones | 39

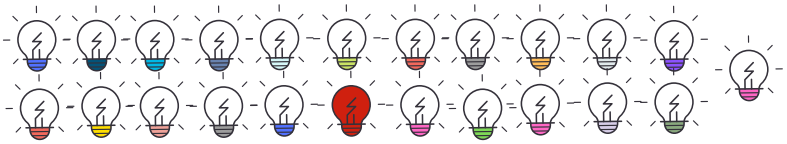
YOU ARE WHAT YOU EAT

Living and eating healthy has never been this easy | 54

What are 17q12 CNVs?

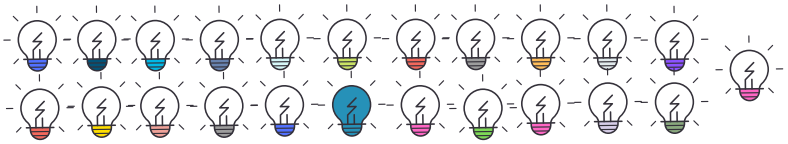
The 17q12 Foundation represents two separate syndromes:

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

in 14500 people

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

in 2500 individuals

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

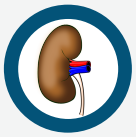
The duplication is most often inherited from a parent. Oftentimes, the duplication is only identified in parents after the child is diagnosed and could have similar, milder or apparently no features

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, 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

How are 17q12 CNVs diagnosed?

BY: MOLLY GOLDMAN & DANIEL MORENO DE LUCA

Now that we know more about 17q12 CNVs, how do people get a diagnosis?

Families arrive at a diagnosis in one of many ways: they may go to see their doctors because of developmental or psychiatric conditions, or because of issues with their kidneys or abnormal blood sugar levels. Doctors can then perform several tests, including genetic testing, where the 17q12 CNVs are revealed.

The main genetic test ordered by doctors in this setting is Chromosomal Microarray. This is a technique looks for small deletions and duplications throughout the genome, and can detect CNVs. In some locations, such as 17q12, these CNVs can be recurrent, meaning that they involve the same unique segment of DNA in unrelated people. The genetic region involved in 17q12 CNVs is about 1.4 Mb in size and includes 15 genes, among them *LHX1* and *HNF1B*. *LHX1* is expressed in the brain early in fetal development and *HNF1B* is important for the developing kidney, liver and pancreas. Children and adults with 17q12 CNVs may share some common features, while some other characteristics could be specific to people with the deletion, or those with the duplication. These features do not always affect everyone with 17q12 CNVs in the same way, and in any given person, they can be more or less obvious. Much remains to be discovered.

In some cases, people with 17q12 CNVs are the first in their families to have this genetic change. When these CNVs are not inherited from either of the biological parents, they are labeled as *de novo*, meaning that they happened spontaneously. Less often, 17q12 CNVs are inherited from a parent. When this happens, the parents appear to be affected most of the time when they have the deletion, and sometimes when they have the duplication, although the degree can be very variable; in some cases these parents are just mildly affected and would go undiagnosed until they are tested to follow up the genetic findings in their children.

Whether the deletion is inherited or *de novo*, as a parent there is nothing you did to cause it and nothing you could have done would have prevented it from occurring in your child. No environmental, dietary, workplace or lifestyle factors are known to cause these changes in 17q12.