How are 17q12 CNVs diagnosed?

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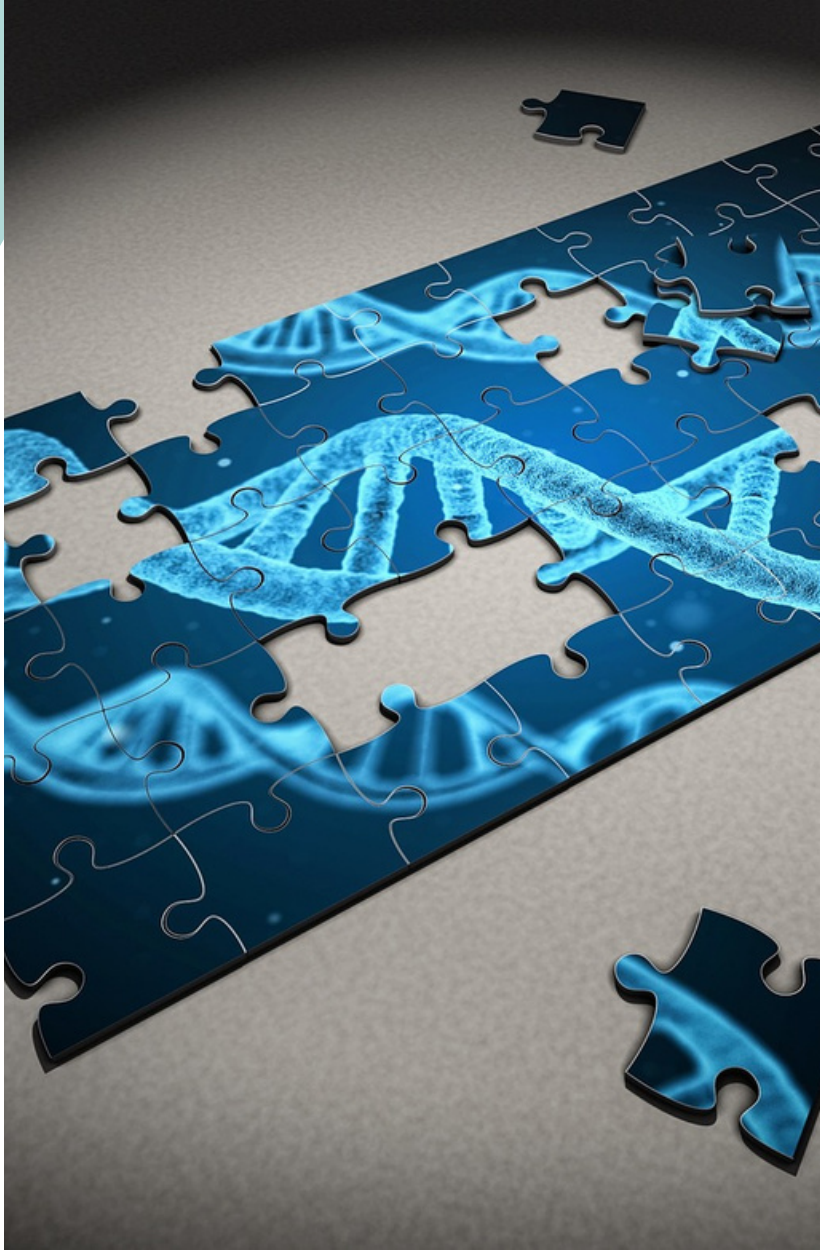
Now that we know more about 17q12 CNVs, how do people get a diagnosis?

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.