SIMONS VIP CONNECT
VARIATION IN INDIVIDUALS PROJECT

16p11.2 Duplication Fact Sheet

What does it mean to have a 16p11.2 Duplication?

A 16p11.2 duplication is a type of genetic change called a *Copy Number Variant (CNV).* A CNV means there is a section of a chromosome missing or extra. Most people have 23 pairs of chromosomes and within those chromosomes there are about 25,000 genes. When a person has a 16p11.2 duplication, typically a group of about 29 genes are repeated, or duplicated (some people may have more or less than this specific number of genes). This means that one chromosome 16 has the expected number of genes, while the other chromosome 16 has extra information.

Researchers now know that this specific section of chromosome 16 contains genes that play an important role in health, development, learning and behavior.

How common is the 16p11.2 Duplication?

- 3 in 1,900 people in the general population have it
- 4 in 10,000 people with a language disorder have it

What are the most common features of the 16p11.2 duplication?

While some people with a 16p11.2 duplication may experience developmental differences as well as medical issues, others have few, if any, concerns. In general, the most common features are:

- Developmental delay of thinking, learning, speech, language and motor skills.
- Behavior concerns including features of Autism Spectrum Disorder and Attention Deficit Hyperactivity Disorder
- Psychiatric conditions including schizophrenia, anxiety, and depression
- Growth patterns of smaller head size (microcephaly), being shorter, or having trouble gaining weight
- Differences in brain structure and function, occasionally including seizures

Not everyone with the duplication will have the same challenges or abilities. Family traits, environmental factors, and other genetic changes all contribute to how a 16p11.2 duplication affects a person. Several scientific articles have been published with results from research about people with this duplication. Summaries of these articles are available on the Simons VIP website.

How is the 16p11.2 duplication inherited?

16p11.2 duplications can be inherited or *de novo.* Many 16p11.2 duplications are found to be *de novo*, meaning that the genetic change is not present in either parent, and is brand new in the child. However, in a number of families, the duplication is passed down from parent to child. If a parent carries the 16p11.2 duplication, then there is a 50% chance of passing it to each child.

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What kind of genetic testing is performed to identify a 16p11.2 duplication?

Most commonly, a 16p11.2 duplication is detected using a genetic test called a *microarray*. This test scans a person's DNA to look for extra or missing sections of the chromosomes.

Another test, called *FISH (fluorescence in situ hybridization)*, can look specifically at the 16p11.2region, and is often used for testing other family members for the same genetic change. .

Are there any management suggestions for individuals with a 16p11.2 duplication?

Anyone found to have a 16p11.2 duplication should be evaluated to see how the duplication has affected them and should continue routine follow-up care. Suggested screening and management includes:

- Routine medical check-ups, including measurement of height and weight, and general review of health
- Developmental assessment with cognitive and behavioral testing, repeated as needed
- Consider consultation with a neurologist and EEG testing if there is a possibility of seizures
- Consider imaging of the spine in patients with spinal curvature or other spine issues
- Consider echocardiogram to examine the heart
- Consider renal ultrasound to examine the kidneys
- Early introduction of speech therapy, if needed

Where can I learn more?

For more information, you can visit the website (www.simonsvipconnect.org) or contact the Simons VIP coordinators at 855-329-5638 or coordinator@simonsvipconnect.org.

You may also want to check out the following resources:

- Unique (www.rarechromo.org)
- Genetics Home Reference (http://ghr.nlm.nih.gov/condition/16p112-duplication)