

## 16p11.2 Deletion Fact Sheet

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

A 16p11.2 deletion 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 deletion, typically a group of about 29 genes are missing, or deleted (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 is missing 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 Deletion?

- 1 in 100 people with autism have it
- 1 in 1,000 people with a language disorder have it
- 1 in 2,300 people in the general population have it

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

While some people with a 16p11.2 deletion may experience developmental differences as well as medical issues, others may 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, Attention Deficit Hyperactivity Disorder, and psychiatric conditions.
- Growth patterns of larger head size (macrocephaly) and being heavier
- Differences in brain structure and function, including seizures.
- Other medical problems including low muscle tone and scoliosis.

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

### How is the 16p11.2 deletion inherited?

16p11.2 deletions can be inherited or *de novo*. The majority of 16p11.2 deletions 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 some families, the deletion is passed down from parent to child. If a parent carries the 16p11.2 deletion, then there is a 50% chance of passing it to each child.

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

Most commonly, a 16p11.2 deletion 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 16p11.2 region, 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 deletion?**

Anyone found to have a 16p11.2 deletion should be evaluated to see how the deletion 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 x-ray 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
- Screening for hypertension and diabetes in patients who are overweight or obese
- Early introduction of speech therapy, if needed

**Where can I learn more?**

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

You may also want to check out the following resources:

- Unique ([www.rarechromo.org](http://www.rarechromo.org))
- Genetics Home Reference (<http://ghr.nlm.nih.gov/condition/16p112-deletion-syndrome>)
- Gene Reviews (<http://www.ncbi.nlm.nih.gov/books/NBK11167/>)