

1q21.1 Deletion Fact Sheet

What does it mean to have a 1q21.1 Deletion?

A 1q21.1 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 1q21.1 deletion, typically a group of about 8 genes are missing, or deleted (some people may have more or less than this specific number of genes). This means that one chromosome 1 has the expected number of genes, while the other chromosome 1 is missing information.

Researchers now know that this specific section of chromosome 1 contains genes that play an important role in health, development, learning and behavior.

How common is the 1q21.1 Deletion?

1q21.1 deletion is a rare CNV; there are no estimates of how many people carry this change yet.

What are the most common features of the 1q21.1 deletion?

While some people with a 1q21.1 deletion may experience developmental differences as well as medical issues, others may have few, if any, concerns. In general, the most common features are:

- Mood and anxiety disorders
- Short stature
- Smaller head size (microcephaly)
- Low muscle tone (hypotonia)
- Other medical problems including cataracts, tremors, and heart problems

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 1q21.1 deletion 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 1q21.1 deletion inherited?

1q21.1 deletions can be *inherited*, meaning that they are passed down from parent to child; or they can be *de novo*, meaning that they are not present in either parent and are brand new in the child. If a parent carries the 1q21.1 deletion, then there is a 50% chance of passing it to each child.

What kind of genetic testing is performed to identify a 1q21.1 deletion?

Most commonly, a 1q21.1 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 1q21.1 region, and is often used for testing other family members for the same genetic change.

Are there any management suggestions for individuals with a 1q21.1 deletion?

A paper published in June 2015 titled "Clinical phenotype of the recurrent 1q21.1 copy-number variant" has much more detailed information about the characteristics seen in people with a 1q21.1 CNV along with medical management suggestions. These suggestions of what the medical evaluation for someone with a 1q21.1 CNV should include are:

1. Psychiatric and neurologic evaluations at several points throughout life: childhood, adolescence, and adulthood.
2. Evaluation by a developmental pediatrician at a young age for autism spectrum disorder (ASD), intellectual disability, attention deficit hyperactivity disorder (ADHD), motor difficulties.
3. Hearing screening as part of well-child visits during childhood, as there were a greater proportion of children with hearing issues
4. Evaluation for both structural and rhythmic heart abnormalities with an echocardiogram and EKG

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/1q211-microdeletion>)
- Gene Reviews (<http://www.ncbi.nlm.nih.gov/books/NBK52787/>)