

## 1q21.1 Duplication Fact Sheet

### What does it mean to have a 1q21.1 Duplication?

A 1q21.1 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 1q21.1 duplication, typically a group of about 8 genes are extra, or duplicated (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 has extra 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 Duplication?

- 3 in 10,000 people in the general population
- 15 or 20 times more common in people with mental health problems or congenital heart disease

### What are the most common features of the 1q21.1 Duplication?

While some people with a 1q21.1 duplication 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
- Speech delay (impairment of phonological processing/articulation) and delayed motor skills
- Behavior concerns including features of autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD)
- Psychiatric conditions including schizophrenia, anxiety, and depression
- Other medical problems including gastric ulcers, scoliosis, low muscle tone (hypotonia), and larger head size (macrocephaly)

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 1q21.1 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 1q21.1 duplication inherited?

1q21.1 duplications 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 duplication, then there is a 50% chance of passing it to each child.

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

Most commonly, a 1q21.1 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 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 duplication?**

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 ASD, intellectual disability, 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 than in children without a 1q21.1 CNV
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](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/1q211-microduplication>)