

# SIMONS SEARCHLIGHT

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## 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 a section of a chromosome is missing or extra. Most people have 23 pairs of chromosomes, and those chromosomes contain about 25,000 genes. A person who has a 1q21.1 duplication has, typically, eight genes that are extra, or duplicated. Some people have more or fewer than eight extra genes. One of the person's copies of chromosome 1 has the expected number of genes, while the other chromosome 1 has extra information.

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

### How common is the 1q21.1 duplication?

- It is found in 3 in 10,000 people in the general population.
- It is 15 to 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 autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD).
- Psychiatric conditions including schizophrenia, anxiety, and depression.
- Other medical problems including stomach ulcers, curved spine (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. Researchers have published scientific articles about how this duplication affects people. Summaries of these articles are available on the Simons Searchlight website.

### Is the 1q21.1 duplication inherited?

Not necessarily. A 1q21.1 duplication can be inherited—that is, passed down from parent to child. Or it can be 'de novo,' meaning that it is not present in either parent and is brand new in the child. If a parent carries the 1q21.1 duplication, there is a 50% chance of passing it down to each child.

### **What kind of gene tests are used to find a 1q21.1 duplication?**

A test called a microarray is most commonly used. This test scans a person's DNA to look for extra or missing sections of chromosomes.

Another test, called FISH (fluorescence in situ hybridization), can look at only the 1q21.1 region. It is often used to test other family members for the same gene change.

### **How should a 1q21.1 duplication be managed?**

A paper published in 2016 titled "[Clinical phenotype of the recurrent 1q21.1 copy-number variant](#)" gives detailed information about the characteristics of people with a 1q21.1 duplication, and also medical management suggestions. The paper suggests that someone with the duplication should receive:

1. Psychiatric and neurologic evaluations in childhood, adolescence, and adulthood.
2. Evaluation by a developmental pediatrician at a young age for ASD, intellectual disability, ADHD, motor difficulties.
3. Hearing tests as part of well-child visits. A greater proportion of children with a 1q21.2 duplication have hearing issues than do children without the duplication.
4. EKG testing for heart problems.

### **Where can I learn more?**

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

You may also find the following resources useful:

- [Unique](#)
- [Genetics Home Reference](#)