1q21.1 deletion syndrome

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Simons Powering Autism Research

SIMONS SEARCHLIGHT
Driven by science. United by hope.

This guide is not meant to take the place of medical advice.

Please consult with your doctor about your genetic results and health care choices. The information in this guide was up to date at the time it was written in 2020. But new information may come to light with new research. You may find it helpful to share this guide with friends and family members or doctors and teachers of the person who has 1q21.1 deletion syndrome.

What is 1q21.1 deletion syndrome?

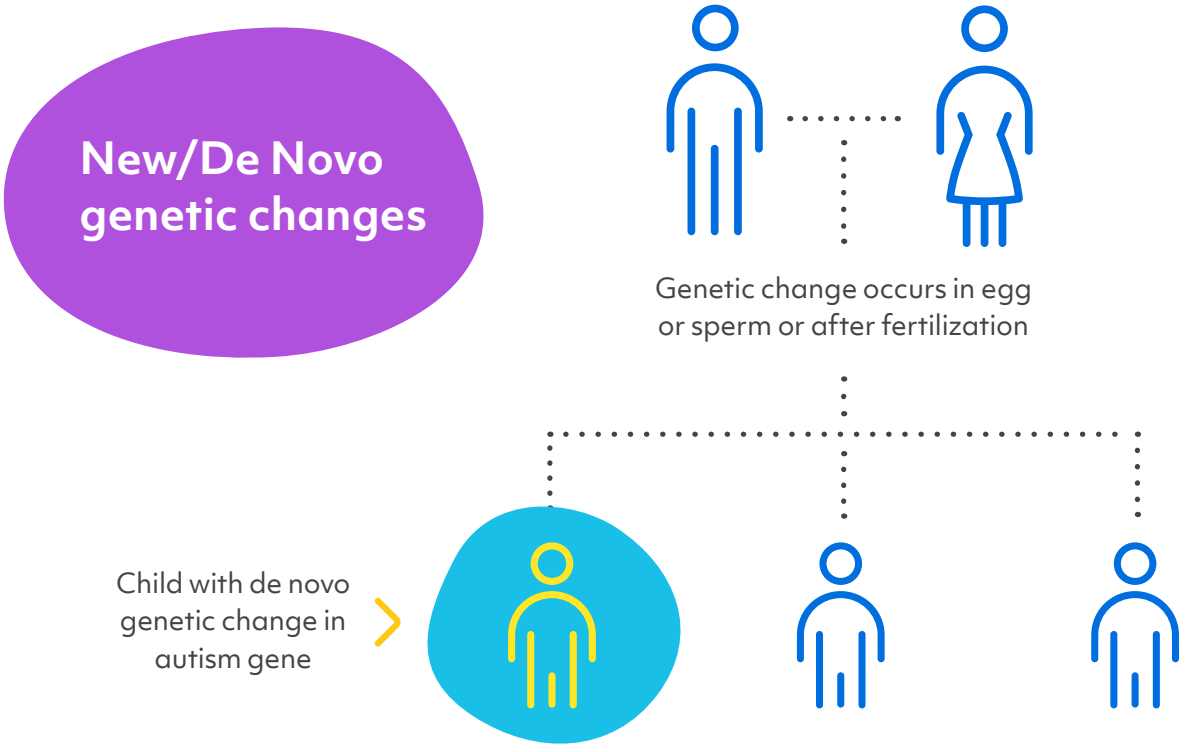
People who have 1q21.1 deletion syndrome may have:

- Developmental delay
- Autism spectrum disorder
- Mood disorders and anxiety
- Short height
- Cataracts, or a clouding in the lens of the eyes that affects vision
- Heart problems

What causes 1q21.1 deletion syndrome?

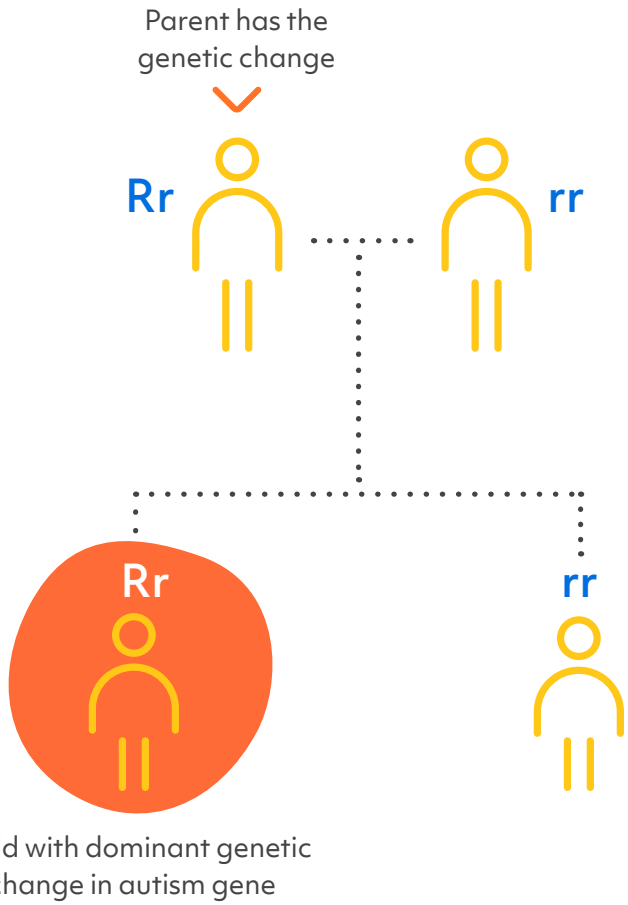
1q21.1 deletion syndrome happens when someone is missing piece of chromosome 1, one of the body's 46 chromosomes. Chromosomes are structures in our cells that house our genes.

We inherit chromosomes from our parents. When the sperm from the father joins the egg from the mother, they form a single cell with 46 chromosomes — 23 from the mother and 23 from the father. This cell then makes many copies of itself.



Dominant inheritance

Children have a 50% chance of inheriting the genetic change



Some people inherit a genetic change from a parent. In other people, small mistakes can occur when genes are being copied. Parts of the chromosomes can break off, make extra copies, or end up in a different order than expected. When this happens, it is called a "de novo", or new, change. The child can be the first in the family to have the genetic change.

Why does my child or I have 1q21.1 deletion syndrome?

No parent causes their child's 1q21.1 deletion syndrome. We know this because no parent has any control over the chromosome changes that they do or do not pass on to their children. Please keep in mind that nothing a parent does before or during the pregnancy causes this to happen. The chromosome change takes place on its own and cannot be predicted or stopped.

What are the chances that other family members or future children will have 1q21.1 deletion syndrome?

Each family is different. A geneticist or genetic counselor can give you advice on the chance that this will happen again in your family.

The risk of having another child who has 1q21.1 deletion syndrome depends on the chromosomes of both birth parents.

- If neither birth parent has the same chromosome change found in their child, the chance of having another child who has the syndrome is on average 1 percent. This 1 percent chance is higher than the chance of the general population. The increase in risk is due to the very unlikely chance that more of the mother's egg cells or the father's sperm cells carry the same change in the gene.
- If one birth parent has the same chromosome change found in their child, the chance of having another child who has the syndrome is about 50 percent.

For a symptom-free sibling, a brother or sister, of someone who has 1q21.1 deletion syndrome, the risk of having a child who has the syndrome depends on the symptom-free sibling's genes and their parents' genes.

- If neither parent has the same chromosome change found in their child who has the syndrome, the symptom-free sibling has a nearly 0 percent chance of having a child who has 1q21.1 deletion syndrome.
- If one birth parent has the same chromosome change found in their child who has the syndrome, the symptom-free sibling has a small chance of also having the same chromosome change.
- If the symptom-free sibling has the same chromosome change as the child who has the syndrome, the symptom-free sibling's chance of having a child who has 1q21.1 deletion syndrome is about 50 percent.

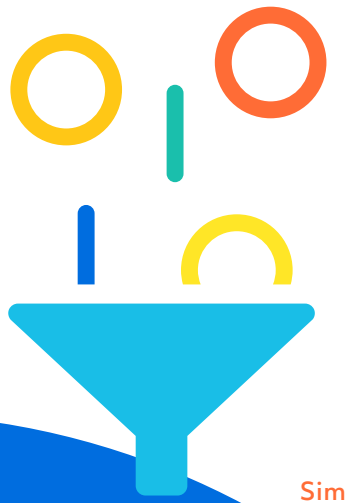
For a person who has 1q21.1 deletion syndrome, the risk of having a child who has the syndrome is about 50 percent.

Do all people who have 1q21.1 deletion syndrome have symptoms?

Not necessarily. Some people do not have any symptoms. Some people may not learn that they have this genetic change until it is found in their children.

Will all of the people in a family that have 1q21.1 deletion syndrome have the same symptoms?

Not necessarily. Family members who have the same chromosome change can have different symptoms.



How many people have 1q21.1 deletion syndrome?

As of 2020, doctors had described a few dozen people who have 1q21.1 deletion syndrome. The first case was found in 2008. There are likely many more undiagnosed people who have the syndrome. Scientists expect to find more people who have the syndrome as access to genetic testing improves.

Do people who have 1q21.1 deletion syndrome look different?

People who have 1q21.1 deletion syndrome may look different. Some people have a small head, also called microcephaly.

How is 1q21.1 deletion syndrome treated?

At this point, there are no medicines designed to treat 1q21.1 deletion syndrome. A genetic diagnosis can help people decide on the best way to track the condition and manage therapies. Doctors can refer people to specialists for

- Physical exams and brain studies
- Genetics consults
- Developmental and behavior studies
- Other issues, as needed

A developmental pediatrician, neurologist, or psychologist can follow progress over time and can help:

- Suggest the right therapies. This can include physical, occupational, speech, or behavioral therapy.
- Guide individualized education plans (IEPs).

Specialists advise that therapies for 1q21.1 deletion syndrome should begin as early as possible, ideally before a child begins school.

If seizures happen, consult a neurologist. There are many types of seizures, and not all types are easy to spot. To learn more, you can refer to resources such as the Epilepsy Foundation's website: epilepsy.com/learn/types-seizures.

1q21.1 deletion syndrome is rare. Doctors and scientists have just recently begun to study it.

This section includes a summary of information from two published articles describing fewer than 30 people. It highlights how many people have different symptoms. See the Sources and references section of this guide for a list of articles.

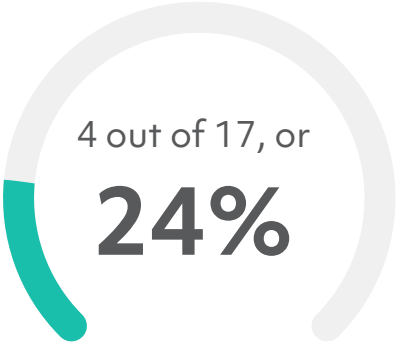
Behavior and development concerns linked to 1q21.1 deletion syndrome

Behavior

Anxiety and mood disorders
5 out of 19 people, or **26%**

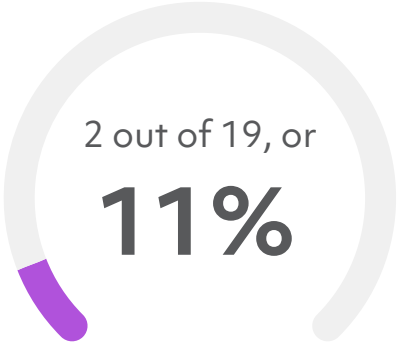
Autism 2 out of 19, or **11%**

Speech



have **difficulty forming certain sounds**

Learning



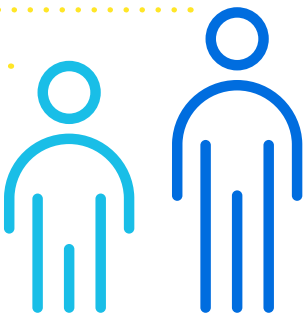
have **intellectual disability**

Medical and physical concerns linked to 1q21.1 deletion syndrome

Growth

Short height
6 out of 12 people, or **50%**

Small head size
4 out of 18 people, or **22%**



Brain and nervous system

Tremor
8 out of 18 people, or **44%**

Seizures
3 out of 17 people, or **18%**

Muscle tone

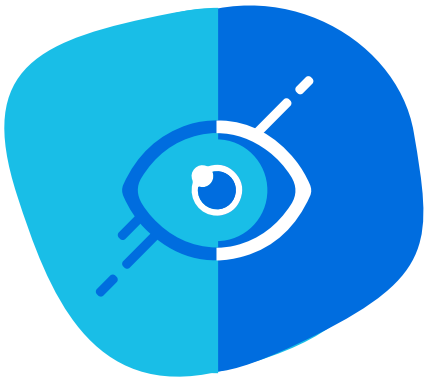
Low muscle tone
7 out of 18 people, or **39%**

Overactive reflexes
Also called hyperreflexia,
6 out of 17 people, or **35%**

Medical and physical concerns linked to 1q21.1 deletion syndrome

Eyes and eyesight

Cataracts, or cloudy areas on the lens of the eyes that can affect vision: 4 out of 12 people, or 33% have cataracts, or cloudy areas on the lens of the eyes that can affect vision.

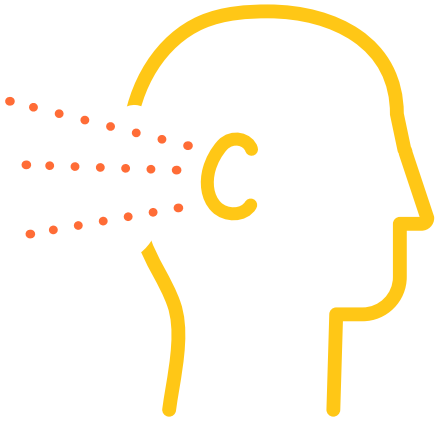


Heart problems

4 out of 12 people, or **33%**

Ears and hearing

Hearing loss **17%**



Simons Searchlight is another research program sponsored and run by the Simons Foundation Autism Research Initiative, also known as SFARI. As part of the next step in your research journey, Simons Searchlight offers you the opportunity to partner with scientists and other families who have the same gene change. Simons Searchlight is a registry for more than 150 genetic changes that are associated with neurodevelopmental conditions, including autism spectrum disorder. Simons Searchlight makes it easier for researchers to access the information they need to advance research on a condition. To register for Simons Searchlight, go to the Simons Searchlight website at www.simonssearchlight.org and click **"Join Us Today"**.

- **Simons Searchlight page on 1q21.1 deletion syndrome**
  www.simonssearchlight.org/research/what-we-study/1q21-1-deletions
- **Simons Searchlight 1q21.1 deletion Facebook community**
  www.facebook.com/groups/168207334043737

Sources and References

The content in this guide comes from published studies on 1q21.1 deletion syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.

- Bernier R. *et al. Genetics in Medicine: Official Journal of the American College of Medical Genetics*, **18**, 341-349, (2016). Clinical phenotype of the recurrent 1q21.1 copy-number variant www.nature.com/articles/gim201578

- Buse M. *et al. Italian Journal of Pediatrics*, **43**, 61, (2017). Expanding the phenotype of reciprocal 1q21.1 deletions and duplications: a case series www.ncbi.nlm.nih.gov/pmc/articles/PMC5518118

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