



# Chromosome 1q21.1 microduplication syndrome





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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 2019. 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 chromosome 1q21.1 microduplication syndrome.



# What is chromosome 1q21.1 microduplication syndrome?



Chromosome 1q21.1 microduplication syndrome can affect communication, social, and learning skills. People who have chromosome 1q21.1 microduplication syndrome may have:

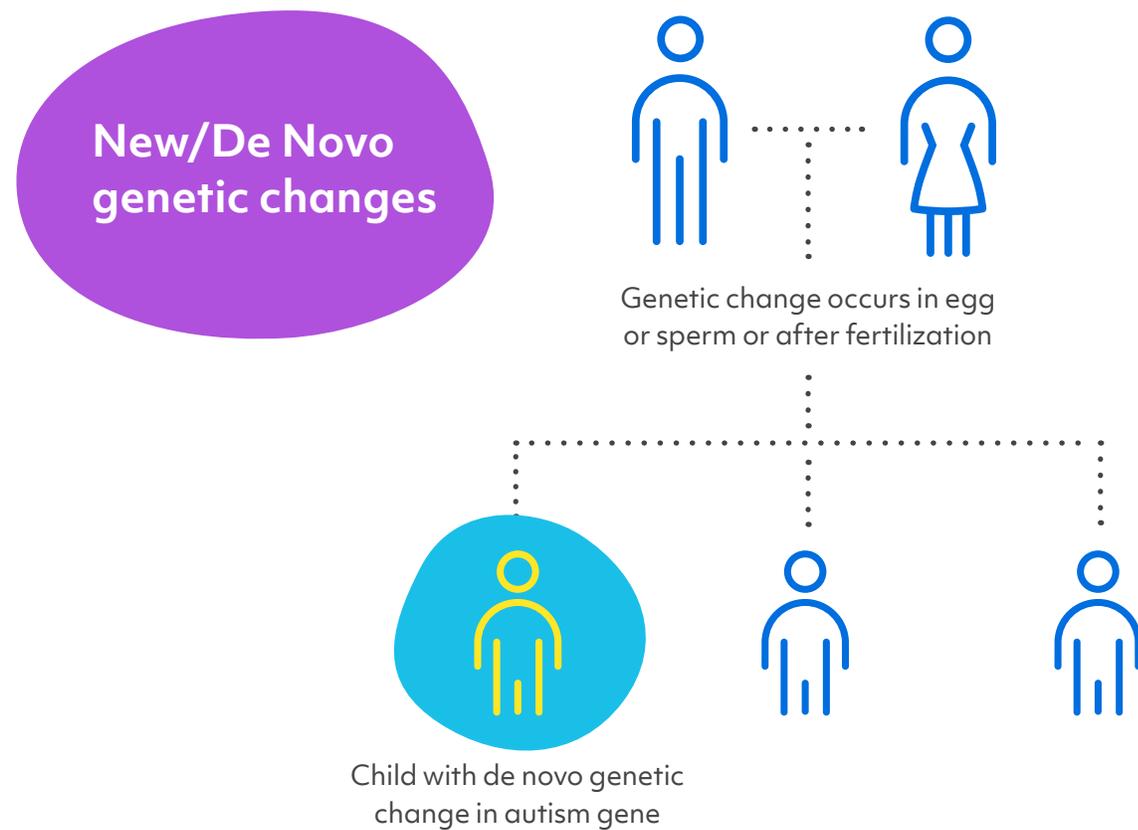
- Developmental delay
- Intellectual disability
- Autism or related behavior issues

# What causes chromosome 1q21.1 microduplication syndrome?

Chromosome 1q21.1 microduplication syndrome is caused when someone has an extra piece of chromosome 1, one of the body's 46 chromosomes.

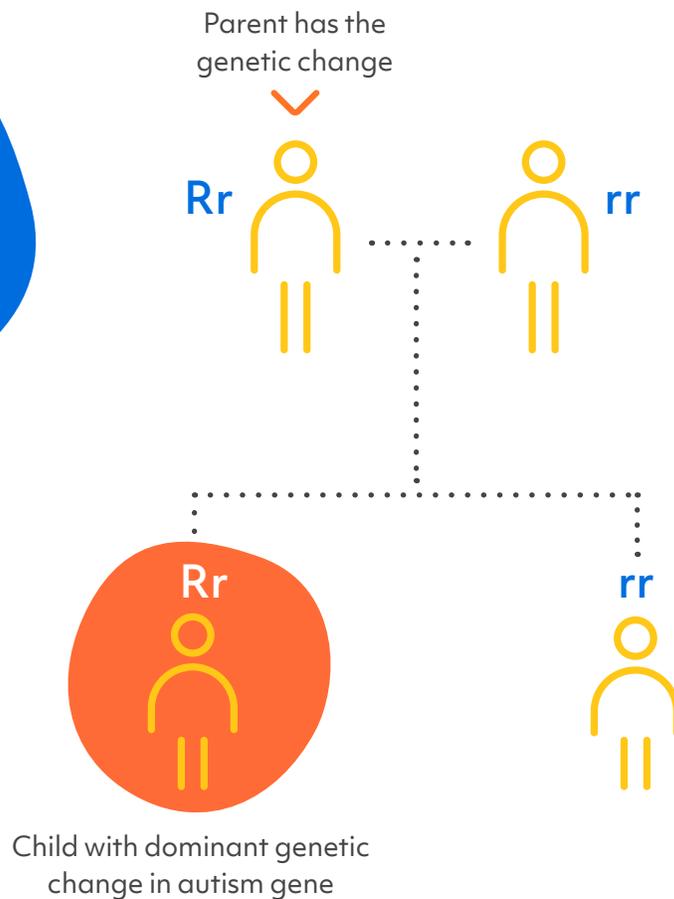
Our cells contain genes that instruct the body on how to grow and function. The genes are arranged in long threads called chromosomes.

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 gene change from a parent. In other people, small mistakes can occur when genes are 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 gene change.

People who have chromosome 1q21.1 microduplication syndrome have an extra piece of chromosome 1. The extra piece can affect learning and how the body develops. Researchers are trying to learn more about what the genes in this extra piece do.

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

No parent causes their child's chromosome 1q21.1 microduplication 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.





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## What are the chances that other family members or future children will have chromosome 1q21.1 microduplication 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 chromosome 1q21.1 microduplication 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 chromosome change.
- 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 chromosome 1q21.1 microduplication 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 the 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 gene 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 chromosome 1q21.1 microduplication syndrome is about 50 percent.

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

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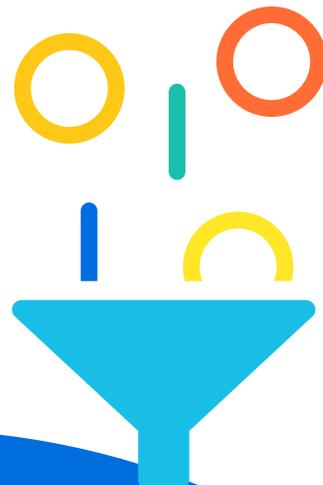
## Do all people who have chromosome 1q21.1 microduplication syndrome have symptoms?

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

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## Do all the people in a family that has the chromosome change have the same symptoms?

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



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## How many people have chromosome 1q21.1 microduplication syndrome?

About 3 in 10,000 people have chromosome 1q21.1 microduplication 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.

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## Do people who have chromosome 1q21.1 microduplication syndrome look different?

In general, people who have chromosome 1q21.1 microduplication syndrome do not look very different.

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## How is chromosome 1q21.1 microduplication syndrome treated?

At this point, there are no medicines designed to treat chromosome 1q21.1 microduplication 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 chromosome 1q21.1 microduplication 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](https://www.epilepsy.com/learn/types-seizures).

Chromosome 1q21.1 microduplication syndrome is rare, found in roughly 3 in 10,000 people.

This section includes a summary of information from published articles. 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 chromosome 1q21.1 microduplication syndrome

### Learning

Many people have learning issues. But most do not have intellectual disability.

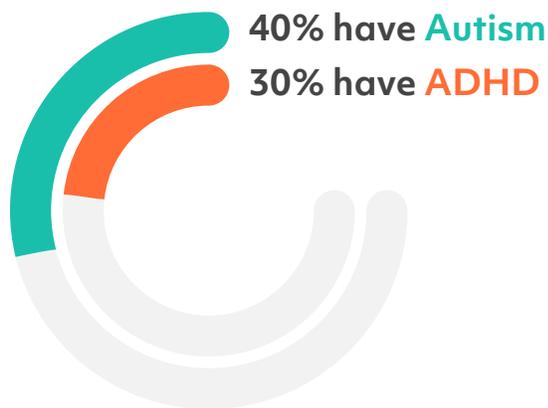
Many people have some level of developmental delay. This often includes issues with fine motor skills, such as using the hands to write.



have **intellectual disability**

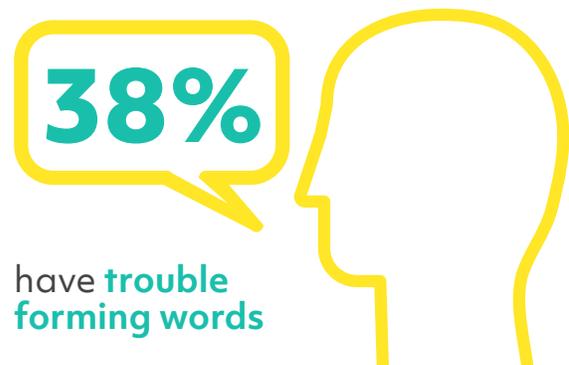
### Behavior

Some people who have the syndrome have attention deficit hyperactivity disorder (ADHD) or autism.

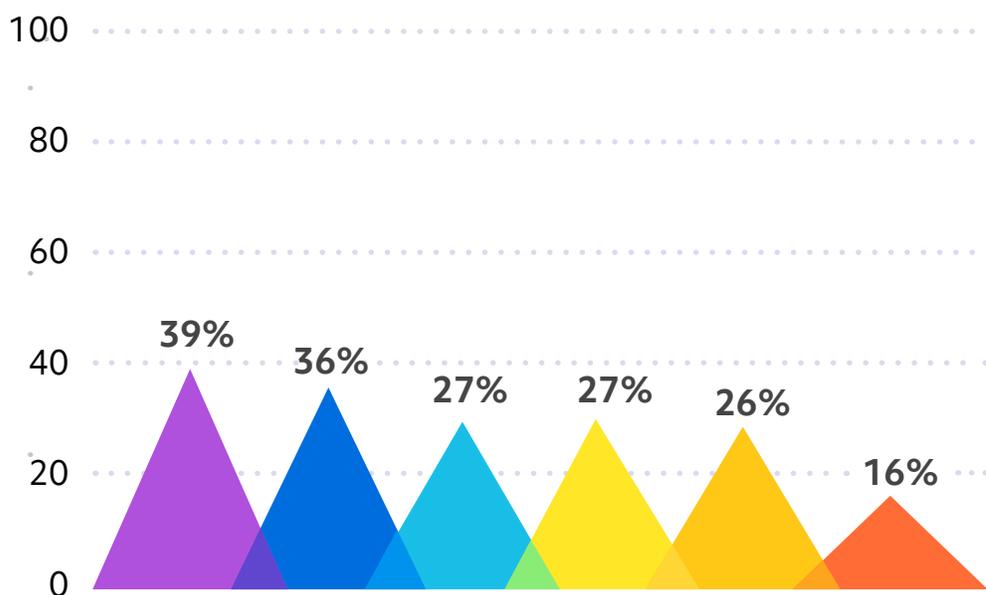


### Speech

Some people have some trouble understanding and forming words.



## Medical and physical concerns linked to chromosome 1q21.1 microduplication syndrome



- Sitting and walking** 4 out of 10 people have trouble walking.
- Joints and spine** About a third have a curved spine, also called scoliosis.
- Growth** More than a quarter are short.
- Eating and gastrointestinal issues** About a quarter of people have stomach ulcers.
- Head** About a quarter have larger than average head size.
- Muscle tone** Almost 2 out of 10 have low muscle tone.



## Where can I find support and resources?

**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](http://www.simonssearchlight.org) and click “Join Us Today”.

- **Learn more about Simons Searchlight**  
[www.simonssearchlight.org/frequently-asked-questions](http://www.simonssearchlight.org/frequently-asked-questions)
- **Simons Searchlight webpage with more information on 1q21.1 microduplication**  
[www.simonssearchlight.org/research/what-we-study/1q21-1-duplications](http://www.simonssearchlight.org/research/what-we-study/1q21-1-duplications)
- **Simons Searchlight Facebook group**  
[www.facebook.com/groups/2081152712152251](https://www.facebook.com/groups/2081152712152251)

## Sources and References

The content in this guide comes from published studies on chromosome 1q21.1 microduplication syndrome. Below you can find details about each study, as well as links to the full articles.

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