Chromosome 15q13.3 microdeletion syndrome
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 15q13.3 microdeletion syndrome.
What is chromosome 15q13.3 microdeletion syndrome?

Chromosome 15q13.3 microdeletion syndrome can affect the development of communication, social, and learning skills. It can affect how a person acts or interacts with others. Many people who have chromosome 15q13.3 microdeletion syndrome have:

- Developmental delay, or intellectual disability, or both
- Autism spectrum disorder or features of autism
- Other behavior issues including attention deficit hyperactivity disorder, also called ADHD, aggression, and rage
- Seizures or abnormal electroencephalogram, also called EEG, results; an electroencephalogram is a test that measures the brain’s electrical activity
- Speech and language delay

The 15q13.3 region on chromosome 15 includes seven genes. Researchers are still trying to learn more about what the genes in this region do. One study suggests that one of the genes deleted in chromosome 15q13.3 microdeletion syndrome, called OTUD7A, is important for brain function.
Chromosome 15q11.2 BP1-BP2 microdeletion syndrome is a rare condition caused when a small piece of DNA is missing from chromosome 15, 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.

**What causes chromosome 15q13.3 microdeletion syndrome?**

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Some people inherit a gene change from a parent. In other people, small mistakes can happen 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 gene change.
Why does my child have chromosome 15q13.3 microdeletion syndrome?

No parent causes their child's chromosome 15q13.3 microdeletion 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 chromosome 15q13.3 microdeletion 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 15q13.3 microdeletion 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 50 percent.

For a symptom-free sibling, a brother or sister, of someone who has chromosome 15q13.3 microdeletion 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 chromosome 15q13.3 microdeletion 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 their sibling who has the syndrome, the symptom-free sibling’s chance of having a child who has chromosome 15q13.3 microdeletion syndrome is 50 percent.

For a person who has chromosome 15q13.3 microdeletion syndrome, the risk of having a child who has the syndrome is about 50 percent.
Do all people who have chromosome 15q13.3 microdeletion 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.

Will all of the people in a family that have chromosome 15q13.3 microdeletion syndrome have the same symptoms?

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

How many people have chromosome 15q13.3 microdeletion syndrome?

As of 2018, doctors had found more than 150 people in the world with chromosome 15q13.3 microdeletion 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.
How is chromosome 15q13.3 microdeletion syndrome treated?

Scientists and doctors have only just begun to study chromosome 15q13.3 microdeletion syndrome. At this point, there are no medicines designed to treat the 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 15q13.3 microdeletion 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.
Chromosome 15q13.3 microdeletion syndrome is very rare. Doctors and scientists have just recently begun to study it. As of 2018, five research studies had been published that describe more than 150 people who have the syndrome.

This section includes a summary of information from major published articles. It highlights how many people have different symptoms. To learn more about the articles, see the Sources and references section of this guide.

Behavior and development concerns linked to chromosome 15q13.3 microdeletion syndrome

**Speech**
Many people have speech impairments.

In a study of 73 children, three out of four had speech problems.

**Behavior**
Many people have autism, features of autism, or behavior issues.

- In a study of 77 people about two-thirds had some behavior issues.
- In a study of 18 people, about one-third had autism.
- In a study of 133 people, about one-tenth had rage.
Behavior and development concerns linked to chromosome 15q13.3 microdeletion syndrome

Learning
Most people show some intellectual disability, ranging from mild to severe. Most people need special educational support.

In a study of 18 people who have chromosome 15q13.3 microdeletion syndrome, the average nonverbal IQ was 60, which is 30 points below average.

Mental health
People who have chromosome 15q13.3 microdeletion syndrome can have hyperactivity and attention issues.

In a study of 133 children, about 10% had attention deficit hyperactivity disorder, also called ADHD.
Medical and physical concerns linked to chromosome 15q13.3 microdeletion syndrome

Brain
About one-quarter of people who have the syndrome have seizures. The age at which people had their first seizure varied. Some had seizures in infancy, others at different points in childhood, and two adults started to have seizures in their 40s. The type of seizure also varied. In some cases, the seizures were difficult to control with medication. One gene within the 15q13.3 region, called CHRNA7, is thought to be responsible for the seizures.

Motor skills
Some people who have chromosome 15q13.3 microdeletion syndrome show delays in the development of hand movement skills, also called fine motor skills, and larger movement skills, also called gross motor skills, such as walking.

1/4 of 86 children and adults studied had seizures are overweight.

2/3 of 16 people who had EEG tests had an abnormal pattern of brain activity.

About 2/5 of 41 people in one study had low muscle tone.
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”.

- Learn more about Simons Searchlight
  www.simonssearchlight.org/frequently-asked-questions

- Simons Searchlight page on 15q13.3
  www.simonssearchlight.org/research/what-we-study/15q13-3-deletion

- Simons Searchlight Community 15q13.3 Facebook group
  www.facebook.com/groups/507629060049202
Sources and References

The content in this guide comes from published studies about chromosome 15q13.3 microdeletion syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.


