16p13.11 deletion syndome

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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 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 16p13.11 deletion syndrome.

What is 16p13.11 syndrome?

16p13.11 deletion syndrome can have moderate to severe effects on communication, social, and learning skills. People who have 16p13.11 deletion syndrome may have:

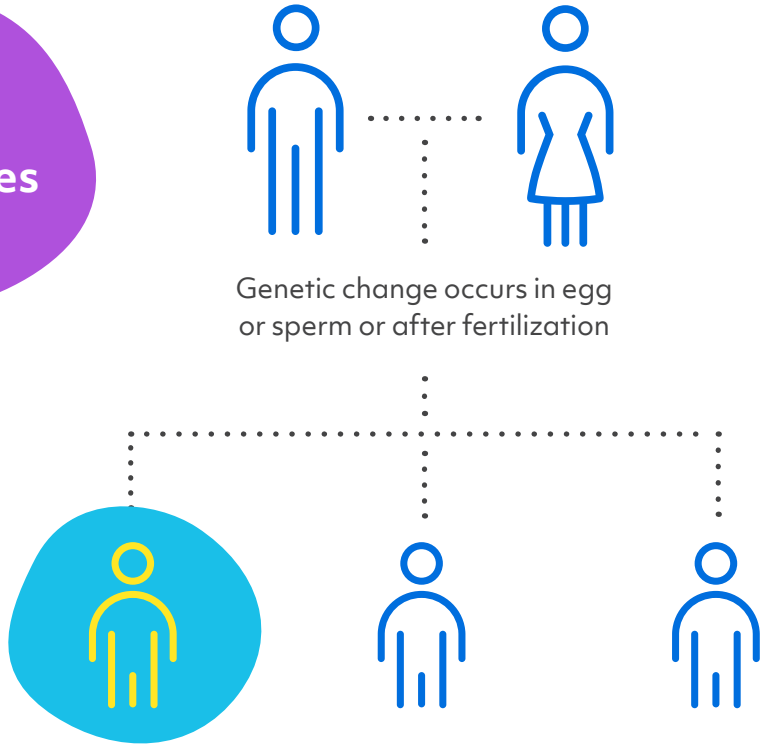
- Intellectual disability
- Autism
- Small head size
- Seizures

What causes 16p13.11 deletion syndrome?

16p13.11 deletion syndrome happens when someone is missing a piece of chromosome 16, one of the body's 46 chromosomes. Chromosomes are structures in our cells that house our genes.

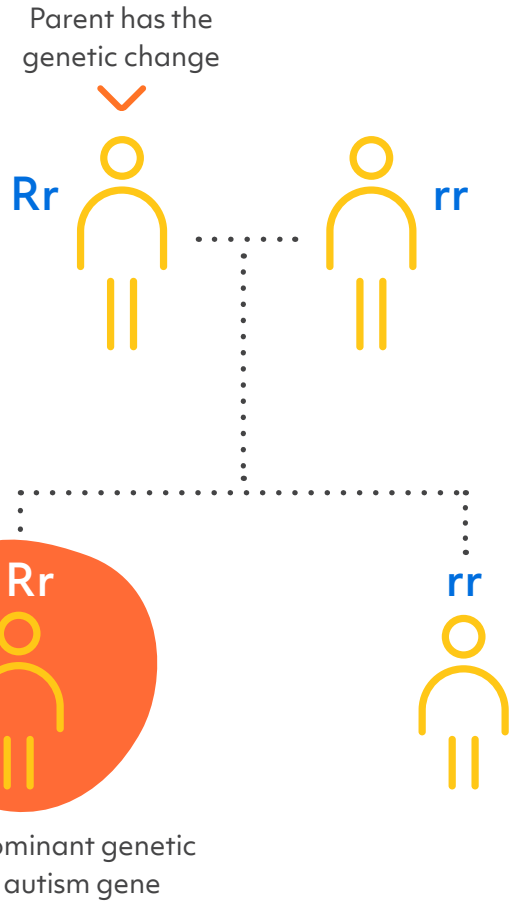
We inherit chromosomes from our parents. When the sperm from the father joins the egg from the mother, they form a single cell with 46 chromosomes — 23 from the mother and 23 from the father. This cell then makes many copies of itself.

New/De Novo genetic changes



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.

Small head size in people who have 16p13.11 deletion syndrome may be due to a gene called NDE1, which sits within the missing region. People who lack two copies of the NDE1 gene have small heads. People who have 16p13.11 deletion syndrome are usually missing one copy of the NDE1 gene.

Why does my child or I have 16p13.11 deletion syndrome?

No parent causes their child's 16p13.11 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 genetic 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 16p13.11 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 16p13.11 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 16p13.11 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 16p13.11 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 16p13.11 deletion syndrome is about 50 percent.

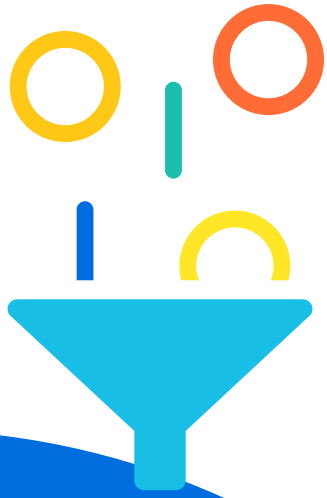
For a person who has 16p13.11 deletion syndrome, the risk of having a child who has the syndrome is about 50 percent.

Do all people who have 16p13.11 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 16p13.11 deletion syndrome have the same symptoms?

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

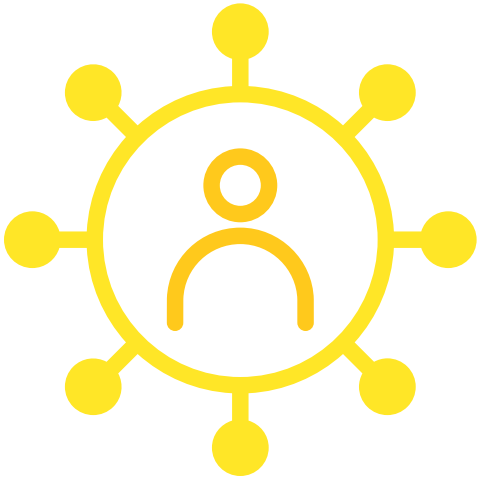


How many people have 16p13.11 deletion syndrome?

As of 2020, doctors had described around 50 people who have 16p13.11 deletion syndrome. The first case was found in 2007. 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 16p13.11 deletion syndrome look different?

People who have 16p13.11 deletion syndrome do not look very different.



How is 16p13.11 deletion syndrome treated?

At this point, there are no medicines designed to treat 16p13.11 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 16p13.11 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.

16p13.11 deletion syndrome is rare, described in about 50 people. Doctors and scientists have just recently begun to study it.

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 16p13.11 deletion syndrome

Learning

Learning problems are common in people who have 16p13.11 deletion syndrome. Combined results from different studies suggest that about 70 percent, or 23/32 people, have intellectual disability.

Speech

Language delay and speech impairment are common.

Behavior

Some people who have 16p13.11 deletion syndrome have autism, obsessive compulsive disorder, or Tourette's syndrome, a disorder with involuntary tics and repeated sounds.

Medical and physical concerns linked to 16p13.11 deletion syndrome

Brain

Seizures are common in people who have 16p13.11 deletion syndrome. Some people have a small head size.

Eyesight

Eyesight issues are common, including crossed eyes.

Sitting and walking

Some people have motor delays.

Muscle tone

Some people have low muscle tone.

Hearing

Some people have hearing issues.

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 and click **"Join Us Today"**.

- **Learn more about Simons Searchlight**
  www.simonssearchlight.org/frequently-asked-questions
- **Simons Searchlight webpage with more information on 16p13-11-deletion**
  www.simonssearchlight.org/research/what-we-study/16p13-11-deletion
- **Simons Searchlight 16p13.11 deletion Facebook Community**
  www.facebook.com/groups/917979968634425

Sources and References

The content in this guide comes from published studies on 16p13.11 deletion syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.

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  www.ncbi.nlm.nih.gov/pmc/articles/PMC2658752
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- Tropeano M. *et al. European Journal of Human Genetics*, **22**, (2014). Clinical utility gene card for: 16p13.11 microdeletion syndrome
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