17q12 deletion 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 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 17q12 deletion syndrome.
What is 17q12 deletion syndrome?

17q12 deletion syndrome can affect communication, social, and learning skills. People who have 17q12 deletion syndrome may have:

- Developmental delay, intellectual disability, or both
- Behavior issues
- Seizures
- Small head, also known as microcephaly
- Brain changes
- Mild changes in facial features
- Kidney issues

Symptoms can vary widely in people who have 17q12 deletion syndrome, even among members of the same family with the same genetic change. Some people may have no noticeable symptoms.
17q12 deletion syndrome is caused when someone is missing a small piece of chromosome 17, 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 17q12 deletion syndrome?

New/De Novo genetic changes

Genetic change occurs in egg or sperm or after fertilization

Child with de novo genetic change in autism gene
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. For 17q12 deletion syndrome, about 70 percent of cases are new, or de novo, and 30 percent are inherited from a parent.
Why does my child have 17q12 deletion syndrome?

No parent causes their child's 17q12 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 17q12 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 17q12 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 17q12 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 17q12 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 17q12 deletion syndrome is about 50 percent.

For a person who has 17q12 deletion syndrome, the risk of having a child who has the syndrome is about 50 percent.
Do all people who have 17q12 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 17q12 deletion syndrome have the same symptoms?

Not necessarily. Family members who have the same chromosome change can have different symptoms.
How many people have 17q12 deletion syndrome?

As of 2019, doctors had found more than 200 people who have 17q12 deletion syndrome. 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 17q12 deletion syndrome look different?

About one-half of those who have 17q12 deletion syndrome have mild physical changes. These can include:

- High, flat forehead
- Low nasal bridge
- Deep-set eyes
- Full cheeks
- High-arched eyebrows
- Small and underdeveloped nails
- Webbing between the second and third fingers, or the second and third toes, or both
- Curved fifth finger
How is 17q12 deletion syndrome treated?

At this point, there are no medicines designed to treat 17q12 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 17q12 deletion syndrome should begin as early as possible, ideally before a child begins school.

Your doctor can recommend whether to see an endocrinologist to check for kidney and urinary issues.

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.
17q12 deletion syndrome is rare — about 200 people have been described in the medical literature. 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. To learn more about the articles, see the Sources and references section of this guide.

**Behavior and development concerns linked to 17q12 deletion syndrome**

**Learning**

About 50%, or 1 in 2 people, of those who have 17q12 deletion syndrome have some degree of developmental delay or learning disability.

About 16%, or 1 in 6 people, have intellectual disability.

**Behavior**

Fewer than 10% of people who have 17q12 deletion syndrome have autism or symptoms of autism.
Medical and physical concerns linked to 17q12 deletion syndrome

**Birth defects**
About **80% to 85%**, or 8 in 10 people, have issues with their kidneys and urinary tract.

**Metabolism**
About **40%**, or 4 in 10 people have a type of diabetes called *maturity-onset diabetes of the young type 5*, or MODY5. MODY5 is most often diagnosed before age 25 years but can range from ages 10 to 50 years.

**Eyes**
About **36%**, or more than 1 in 3 people who have 17q12 deletion syndrome have issues with their eyes, including crossed eyes, uncontrolled movements of the eyes, clouding of the lens, also called cataracts, and eye changes called colobomas, which lead to a hole in one of the structures of the eyes.
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 page on 17q12 deletion syndrome [www.simonssearchlight.org/research/what-we-study/17q12-deletion](http://www.simonssearchlight.org/research/what-we-study/17q12-deletion)
- Simons Searchlight 17q12 deletion Facebook Community [www.facebook.com/groups/235285544024090](http://www.facebook.com/groups/235285544024090)
Sources and References

The content in this guide comes from published studies about 17q12 deletion syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.

- Laffargue F. et al. Archives of Disease in Childhood, 100, 259-264, (2015). Towards a new point of view on the phenotype of patients with a 17q12 microdeletion syndrome
  www.ncbi.nlm.nih.gov/pubmed/20154674

  www.ncbi.nlm.nih.gov/pubmed/27409573

  www.ncbi.nlm.nih.gov/books/NBK401562