7q11.23 duplication syndrome
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 7q11.23 duplication syndrome.

This guide is not meant to take the place of medical advice.
What is 7q11.23 duplication syndrome?

7q11.23 duplication syndrome can affect communication, social, and learning skills. People who have 7q11.23 duplication syndrome may have:

- Distinct facial features
- Speech delay
- Behavior issues, including anxiety, attention deficit hyperactivity disorder, and autism
- Developmental delay
7q11.23 duplication syndrome happens when someone has an extra piece of chromosome 7, 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 7q11.23 duplication 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.

**Dominant inheritance**
Children have a 50% chance of inheriting the genetic change

Parent has the genetic change

Rr  rr

Children have a 50% chance of inheriting the genetic change

Child with dominant genetic change in autism gene

Rr  rr
Why does my child or I have 7q11.23 duplication syndrome?

No parent causes their child's 7q11.23 duplication 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 7q11.23 duplication 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 7q11.23 duplication 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 7q11.23 duplication 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 7q11.23 duplication 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 7q11.23 duplication syndrome is about 50 percent.

For a person who has 7q11.23 duplication syndrome, the risk of having a child who has the syndrome is about 50 percent.
Do all people who have 7q11.23 duplication 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 7q11.23 duplication syndrome have the same symptoms?

Not necessarily. Family members who have the same chromosome change can have different symptoms.
How many people have 7q11.23 duplication syndrome?

About 1 in 7,500 to 1 in 20,000 people have 7q11.23 duplication syndrome. The first case was described 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 7q11.23 duplication syndrome look different?

People who have 7q11.23 duplication syndrome may look different. Appearance can vary and can include some but not all of these features:

- Wide, flat head shape
- Wide forehead
- Straight eyebrows
- Deep-set eyes
- Long eyelashes
- Wide-tipped nose
How is 7q11.23 duplication syndrome treated?

At this point, there are no medicines designed to treat 7q11.23 duplication 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 7q11.23 duplication 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: [www.epilepsy.com/learn/types-seizures](http://www.epilepsy.com/learn/types-seizures).
Most people who have 7q11.23 duplication syndrome have developmental delay, including delayed motor, speech, and social skills as young children. For most children, intellectual ability is low to average. About 20 percent have borderline intellectual disability and 18 percent have intellectual disability.

7q11.23 duplication syndrome is rare, found in roughly 1 in 7,500 to 1 in 20,000 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 7q11.23 duplication syndrome**

**Learning**

Most people who have 7q11.23 duplication syndrome have developmental delay, including delayed motor, speech, and social skills as young children. For most children, intellectual ability is low to average. About 20 percent have borderline intellectual disability and 18 percent have intellectual disability.

**Speech**

Almost all young children who have 7q11.23 duplication syndrome have speech delay or other speech issues, including motor challenges that affect speech. Expressive language, the use of words and gestures to communicate, is usually more delayed than receptive language, the ability to understand information.
Behavior and development concerns linked to 7q11.23 duplication syndrome

- More than one-half have **anxiety issues**.
- More than one-third have **attention deficient hyperactivity disorder**, also called ADHD. (35 percent)
- Almost one-third have **selective mutism**, difficulty speaking in select social settings, such as in school. (29 percent)
- One-quarter have **oppositional disorders**, defiant and disobedient behavior to authority figures. (25 percent)
- About one-fifth have **autism**.
Medical and physical concerns linked to 7q11.23 duplication syndrome

**Movement**
Almost three-quarters of people who have 7q11.23 duplication syndrome have difficulty or delay in learning motor skills, also called developmental coordination disorder.

74%

**Head**
One-half have a large head, known as macrocephaly.

50%

**Muscle tone**
Some people have low muscle tone.

60%

**Sitting and walking**
Some have differences in how they walk or difficulty balancing.

62%

**Heart**
Almost one-half have heart issues, most often a large artery, also known as dilation of the ascending aorta.

46%

**Brain**
Almost one-fifth have seizures.

19%
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 200 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 7q11.23 duplication [www.simonssearchlight.org/research/what-we-study/7q11-23-duplication](http://www.simonssearchlight.org/research/what-we-study/7q11-23-duplication)
- Simons Searchlight 7q11.23 duplication Facebook community [www.facebook.com/groups/729341430825065/about](http://www.facebook.com/groups/729341430825065/about)
Sources and References

The content in this guide comes from published studies on 7q11.23 duplication syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.

- Sanders S.J. et al. *Neuron*, 70, 863-885, (2011). Multiple recurrent de novo copy number variations (CNVs), including duplications of the 7q11.23 Williams-Beuren syndrome region, are strongly associated with autism. [www.ncbi.nlm.nih.gov/pmc/articles/PMC3939065](www.ncbi.nlm.nih.gov/pmc/articles/PMC3939065)
