22q11.2 duplication 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 22q11.2 duplication syndrome.
What is 22q11.2 duplication syndrome?

22q11.2 duplication syndrome can have variable effects on the development of communication, social, and learning skills. It can affect how a person acts or interacts with others. Many people who have 22q11.2 duplication syndrome have:

- Autism spectrum disorder
- Learning disabilities
- Growth delays
- Motor delays
What causes 22q11.2 duplication syndrome?

22q11.2 duplication syndrome is caused when someone has an extra piece of chromosome 22, 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.

New/De Novo genetic changes

Genetic change occurs in egg or sperm or after fertilization

Child with de novo genetic change in autism gene
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.

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 or I have 22q11.2 duplication syndrome?

No parent causes their child’s 22q11.2 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 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 22q11.2 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 22q11.2 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 50 percent.

For a symptom-free sibling, a brother or sister, of someone who has 22q11.2 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 22q11.2 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 their sibling who has the syndrome, the symptom-free sibling’s chance of having a child who has 22q11.2 duplication syndrome is 50 percent.

For a person who has 22q11.2 duplication syndrome, the risk of having a child who has the syndrome is about 50 percent.
Do all people who have 22q11.2 duplication 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 22q11.2 duplication syndrome have the same symptoms?

Not necessarily. Family members who have the same chromosome change can have different symptoms.
How many people have 22q11.2 duplication syndrome?

About 1 in 4,000 newborns have 22q11.2 duplication syndrome. As of 2019, doctors had described about 100 people in the world with 22q11.2 duplication syndrome. The first case was found in 2003. 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 22q11.2 duplication syndrome look different?

Some people who have 22q11.2 duplication syndrome have minor changes in their appearance. Some people have a large head.
How is 22q11.2 duplication syndrome treated?

Scientists and doctors have only just begun to study 22q11.2 duplication 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 22q11.2 duplication syndrome should begin as early as possible, ideally before a child begins school.

If seizures happen, consult a neurologist. There are many different 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.
22q11.2 duplication syndrome is rare. Doctors and scientists have just recently begun to study it. As of 2019, studies found about 100 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 22q11.2 duplication syndrome

Learning

Most people who have 22q11.2 duplication syndrome have mild to moderate intellectual disability.

Behavior

About 70% of people who have 22q11.2 duplication syndrome are either sensitive or insensitive to sensory stimuli, such as bright lights or loud sounds. About 40% have autism.
Medical and physical concerns linked to 22q11.2 duplication syndrome

**Growth**
Many people who have 22q11.2 duplication syndrome have growth delays or are short in height.

**Motor concerns**
Many people have motor delays.

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

**Brain**
About 15% of people who have the syndrome have seizures.

**Ears and hearing**
One small study showed mild hearing impairment in one-third of people who have the syndrome.

**Other issues**
Some people who have the syndrome have problems with their immune system. They may be likely to have infections and unusual reactions to certain vaccines.
Where can I find support and resources?

The International 22q11.2 Foundation
www.22q.org
Sources and References

The content in this guide comes from published studies about 22q11.2 duplication syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.


