



Chromosome 8p23.1 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 chromosome 8p23.1 duplication syndrome.



What is chromosome 8p23.1 duplication syndrome?

Chromosome 8p23.1 duplication syndrome can affect the development of communication, social, and learning skills. It can affect how a person acts or interacts with others. People who have chromosome 8p23.1 duplication syndrome have:

- Developmental delay
- Learning issues



What causes chromosome 8p23.1 duplication syndrome?

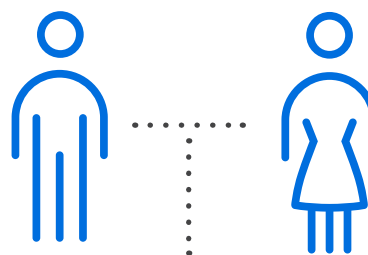
Chromosome 8p23.1 duplication syndrome is caused when someone has an extra piece of chromosome 8, one of the body's 46 chromosomes.

Our cells contain genes that instruct the body on how to grow and function. The genes are arranged in long threads called chromosomes.

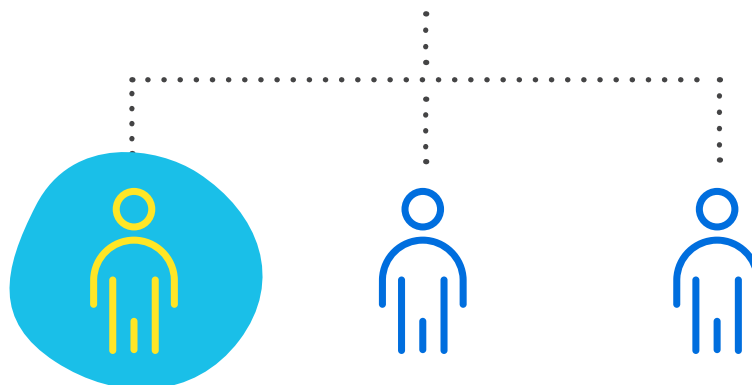
We inherit chromosomes from our parents. When the sperm from the father joins the egg from the mother, they form a single cell with 46 chromosomes — 23 from the mother and 23 from the father. This cell then makes many copies of itself.



New/De Novo
genetic changes



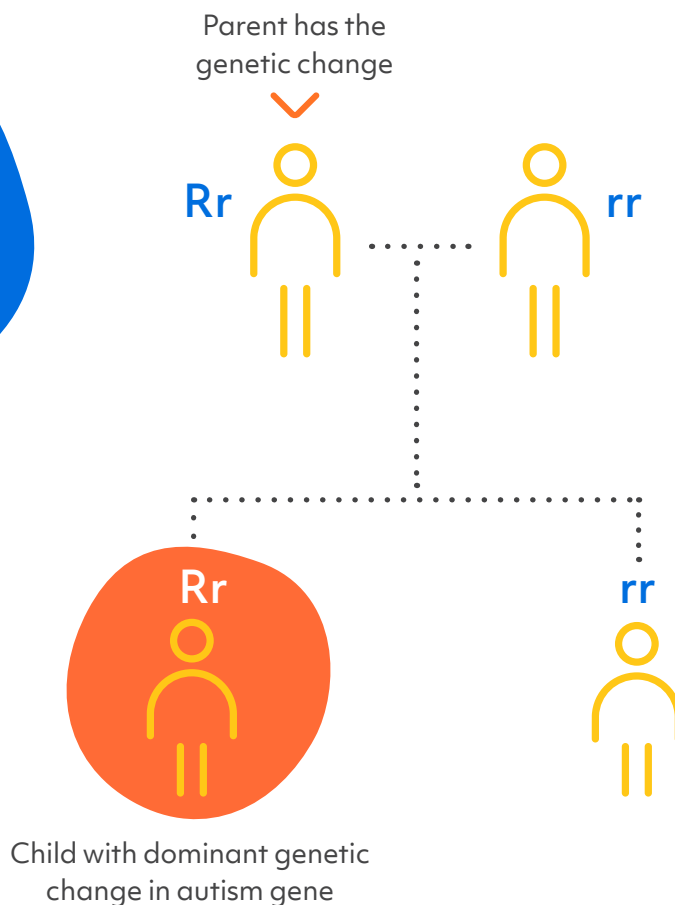
Genetic change occurs in egg
or sperm or after fertilization



Child with de novo genetic
change in autism gene

Dominant inheritance

Children have a 50% chance of inheriting the genetic change




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.

People who have chromosome 8p23.1 duplication syndrome have an extra piece of chromosome 8. The extra piece can affect learning and how the body develops. Researchers are trying to learn more about what the genes in this extra piece do.

Why does my child or I have chromosome 8p23.1 duplication syndrome?

No parent causes their child's chromosome 8p23.1 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 chromosome 8p23.1 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 chromosome 8p23.1 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 chromosome 8p23.1 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 chromosome 8p23.1 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 chromosome 8p23.1 duplication syndrome is 50 percent.

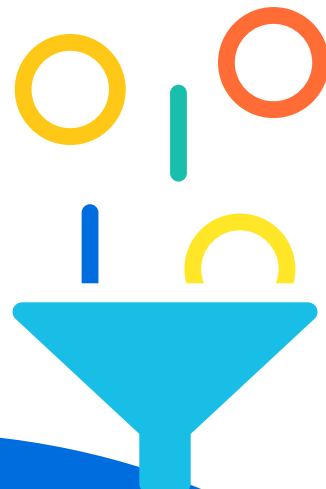
For a person who has chromosome 8p23.1 duplication syndrome, the risk of having a child who has the syndrome is about 50 percent.

Do all people who have chromosome 8p23.1 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 chromosome 8p23.1 duplication syndrome have the same symptoms?

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



How many people have chromosome 8p23.1 duplication syndrome?

A study published in 2015 described 24 people who have chromosome 8p23.1 duplication 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.

Do people who have chromosome 8p23.1 duplication syndrome look different?

People who have chromosome 8p23.1 duplication syndrome may look different from others. About one in four has a cleft lip or cleft palate, which is an opening or split in the upper lip or roof of the mouth that happens before birth. About one in five have changes in the appearance or function of the eyes.



How is chromosome 8p23.1 duplication syndrome treated?

Scientists and doctors have only just begun to study chromosome 8p23.1 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 chromosome 8p23.1 duplication syndrome should begin as early as possible, ideally before a child begins school.

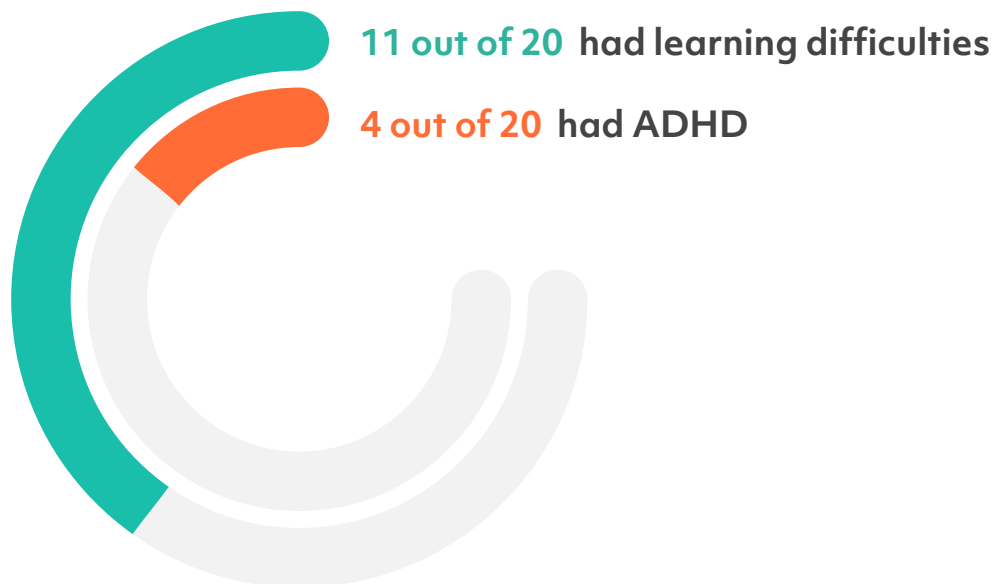
Chromosome 8p23.1 duplication syndrome is very rare. Doctors and scientists have just recently begun to study it. As of 2015, studies found just 24 people who have the syndrome.

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 8p23.1 duplication syndrome

Learning

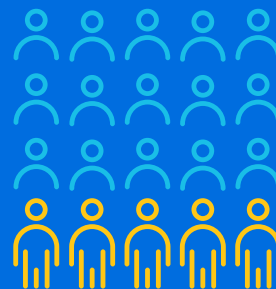
In a study of 20 people who have chromosome 8p23.1 duplication syndrome, about one-half had **learning difficulties**. Nearly one-quarter had attention deficit hyperactivity disorder, also called **ADHD**.



Medical and physical concerns linked to chromosome 8p23.1 duplication syndrome

Muscle tone

In a study of 20 people who have chromosome 8p23.1 duplication syndrome, one-quarter had **low muscle tone**.



5 out of 20 people
had low muscle tone

Ears and hearing

10%

had **hearing loss**
or **sensitivity**.

Birth defects

In a study of 24 people who have the syndrome,

25%

had **heart disease**
at birth.



**Where can I
find support
and resources?**

8p23.1 duplication Facebook groups

www.facebook.com/8p231-DeletionDuplication-Syndrome-611521165649719

www.facebook.com/groups/939907672688019



Project 8P

www.project8p.org

Sources and References

The content in this guide comes from published studies about chromosome 8p23.1 duplication syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.

- Weber A. *et al. Molecular Cytogenetics*, **7**, 94, (2014). 8p23.1 duplication syndrome: Narrowing of critical interval to 1.80 Mbp www.ncbi.nlm.nih.gov/pubmed/25520754
- Barber JC. **et al. American Journal of Medical Genetics Part A**, **167A**, 2052-2064, (2015). Inside the 8p23.1 duplication syndrome; eight microduplications of likely or uncertain clinical significance www.ncbi.nlm.nih.gov/pubmed/26097203



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