17p11.2 duplication syndrome
- or -
Potocki-Lupski 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 Potocki-Lupski syndrome, also called 17p11.2 duplication syndrome.
Potocki-Lupski syndrome affects communication, social, and learning skills. Potocki-Lupski syndrome is also known as 17p11.2 duplication syndrome. People who have Potocki-Lupski syndrome may have:

- Developmental delay
- Intellectual disability
- Attention issues
- Autism
- Low muscle tone
- Difficulty swallowing
- Failure to thrive
- Heart issues
What causes Potocki-Lupski syndrome?

Potocki-Lupski syndrome is caused by an extra 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.

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.
Why does my child or I have Potocki-Lupski syndrome?

No parent causes their child’s Potocki-Lupski 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.
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 Potocki-Lupski 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 Potocki-Lupski 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 Potocki-Lupski 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 Potocki-Lupski syndrome is about 50 percent.

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

Not necessarily. Family members who have the same chromosome change can have different symptoms.
How many people have Potocki-Lupski syndrome?

About 1 in 25,000 people have Potocki-Lupski 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 Potocki-Lupski syndrome look different?

People who have Potocki-Lupski syndrome may have slight changes in their facial features.
How is Potocki-Lupski syndrome treated?

At this point, there are no medicines designed to treat Potocki-Lupski 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

Physicians may monitor people who have Potocki-Lupski syndrome for slow growth, short height, and failure to thrive. Some people who have Potocki-Lupski syndrome are born with heart issues and may need to be monitored by a cardiac specialist.

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 Potocki-Lupski 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.
Potocki-Lupski syndrome/17p11.2 duplication syndrome is rare, found in roughly 1 in 25,000 people.

This section includes a summary of information from a published article. It highlights how many people have different symptoms. To learn more about the article, see the Sources and references section of this guide.

Behavior and development concerns linked to Potocki-Lupski syndrome/17p11.2 duplication syndrome

**Speech**  
Almost everyone who has the syndrome has speech delay. Speech skills may improve with age and speech therapy.

**Learning**  
Most people have moderate intellectual disability.

**Behavior**  
Attention issues, hyperactivity, and anxiety are common. Some people who have the syndrome have autism. One small study estimated that about 60 percent of people have autism.
Medical and physical concerns linked to Potocki-Lupski syndrome/17p11.2 duplication syndrome

Sitting and walking
Most people who have the syndrome have motor delays. Most can walk on their own by 2 years old.

Muscle tone
Low muscle tone is common.

Feeding and digestion issues
Difficulty feeding and swallowing are common.

Growth
Slow growth is common and may be linked to difficulty feeding. Some people have short height and lower than usual levels of growth hormone.

Heart
About 40 percent have heart issues, which are often present at birth. This can include atrial and ventral septal defects, a hole in the wall between the two upper or lower chambers of the heart. A few people have problems with heart rhythms that can be detected with a medical test called an electrocardiogram.

Sleep
Some people have breathing issues during sleep, including sleep apnea, in which breathing stops briefly during sleep.
Where can I find support and resources?

PTLS Foundation
www.ptlsfoundation.org
Sources and References

The content in this guide comes from a published article about 17p11.2 duplication syndrome. Below you can find details about the article, as well as a link to the full article.

  www.ncbi.nlm.nih.gov/books/NBK447920/#potocki-lupski