Duplication 15q 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 duplication 15q syndrome.
What is Duplication 15q syndrome?

Duplication 15q syndrome, also called Dup15q syndrome, can affect communication, social, and learning skills. People who have Dup15q syndrome may have:

- Developmental delay
- Cognitive impairment
- Speech and language delays
- Autism
- Seizures
- Behavior changes
- Mild to moderate physical changes
What causes Dup15q syndrome?

Dup15q syndrome refers to a broad set of conditions that are caused by an extra piece of chromosome 15, one of the body’s 46 chromosomes. Chromosomes are structures in our cells that house our genes. This extra piece happens on a specific part of the chromosome called 15q11.2-q13.1.

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 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.
In Dup15q syndrome, the extra piece of chromosome 15 can happen in two different ways. In about 20% of cases, the extra piece is located within one of the copies of chromosome 15. This is called an interstitial duplication and is also known as int dup(15). In these cases, people have 46 chromosomes, one pair of each of the 23 chromosomes. About 85% of interstitial duplications are new, or de novo. About 15% are inherited from the mother.

Dup15q syndrome can also be caused by a genetic change that leads to a small extra chromosome. People who have this type of Dup15q syndrome have 47 chromosomes, including 3 copies of chromosome 15 — two typical copies and one small extra piece. This is called isodicentric chromosome 15 and is also known as idic(15). This type of Dup15q syndrome is almost always new, or de novo.
Why does my child have Dup15q syndrome?

No parent causes their child’s Dup15q 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 Dup15q 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 Dup15q 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 Dup15q 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 Dup15q 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 Dup15q syndrome is about 50 percent.

For a person who has Dup15q syndrome, the risk of having a child who has the syndrome is about 50 percent.
Do all people who have Dup15q 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 Dup15q syndrome have the same symptoms?

Not necessarily. Family members who have the same chromosome change can have different symptoms.
How many people have Dup15q syndrome?

Scientists don’t know exactly how many people have Dup15q syndrome. But they estimate that it is found in roughly 1 in 5,000 people. In people who have autism, about 1 in 250 to 1 in 500 people have 15q duplications. The first case was found in 1991. 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 Dup15q syndrome look different?

People who have Dup15q syndrome may look different. Appearance can vary and can include some of these features, which are often mild:

- Flattened nasal bridge with a short upturned nose and upward point to the nostrils
- Small jaw
- Low-set ears
- Low-set forehead
- High-arched roof of the mouth
- Full lips
How is Dup15q syndrome treated?

At this point, there are no medicines designed to treat Dup15q 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 Dup15q 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.
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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 Dup15q syndrome

**Learning**
Most people who have Dup15q syndrome have developmental delay and moderate to severe intellectual disability. However, there is some variability. People who have the interstitial duplication are often more mildly affected.

**Behavior**
Most people who have Dup15q syndrome have autism. Of the roughly 20 chromosome changes that have been linked to autism, Dup15q has the highest association with autism.

**Speech**
Most people who have Dup15q syndrome have issues with speech and language, ranging from moderate to severe. Some repeat words, known as echolalia, make repetitive sounds, known as stereotyped utterances, or reverse pronouns. Others may have a hard time learning to make certain speech sounds.

**Mental health**
Some research links the idic(15) form of Dup 15q syndrome to schizophrenia, a mental condition that may include hallucinations and other symptoms. However, it’s unclear if people who have Dup15q syndrome have psychosis, a symptom of schizophrenia in which people lose touch with reality. It can be difficult to detect psychosis in people who have intellectual disability and limited verbal skills.
Medical and physical concerns linked to Dup15q syndrome

Brain

More than one-half of those who have Dup15q syndrome have epilepsy. Seizures usually begin between the ages of 6 months and 9 years and can include different types:

- Infantile spasms: seizures in which the muscles in the arms and legs become stiff, often bending the body forward
- Myoclonic: brief jerks of a muscle or group of muscles
- Tonic-clonic: a seizure that includes a combination of stiffening and jerking
- Absence: seizures with brief staring spells
- Focal: seizures that begin on one side of the brain

A small number of people who have Dup15q syndrome have died from sudden unexpected death in epilepsy, also called SUDEP. These deaths almost always happen during sleep and most, though not all, happen in teenagers and young adults.

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Where can I find support and resources?

Dup15q Alliance
www.dup15q.org
Sources and References

The content in this guide comes from published studies about Dup15q syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.

  www.ncbi.nlm.nih.gov/pubmed/2055888

- Finucane BM. et al. Gene Reviews, (2016). 15q duplication syndrome and related disorders,
  www.ncbi.nlm.nih.gov/books/NBK367946