DYRK1A-Related Syndrome

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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 DYRK1A-related syndrome.

What is DYRK1A-related syndrome?

DYRK1A-related syndrome happens when there are changes to the DYRK1A gene. These changes can keep the gene from working as it should.

Key role

The DYRK1A gene plays a key role in brain development. It is especially important for creating new brain cells and for updating connections among brain cells.

Symptoms

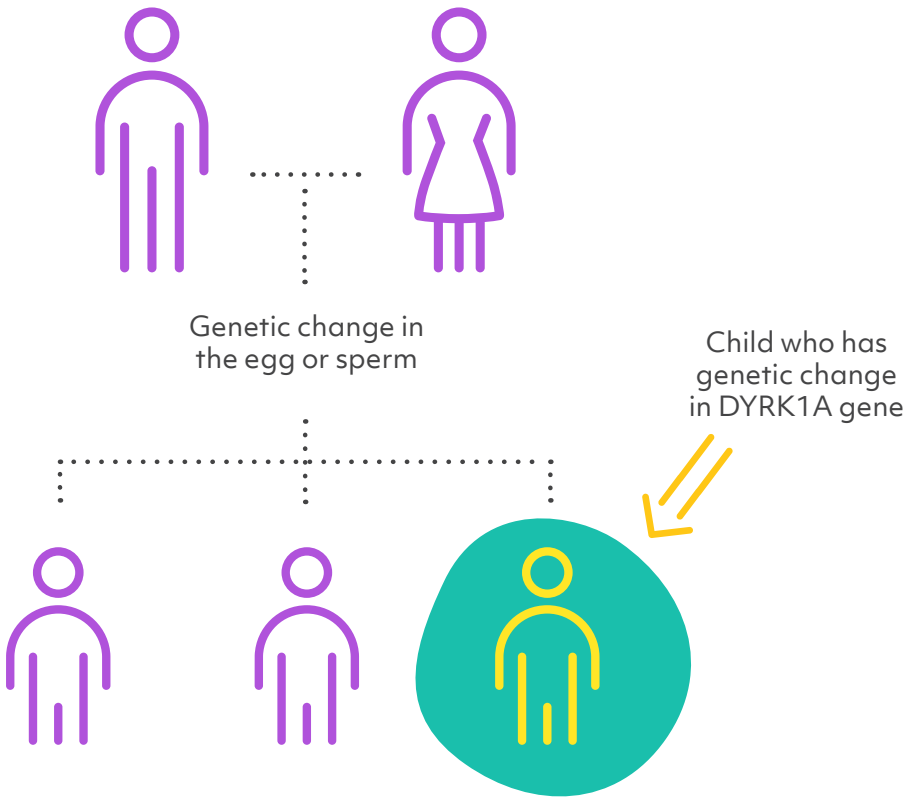
Because the DYRK1A gene is important in brain development, many people who have DYRK1A-related syndrome have:

- Intellectual disability
- Speech delay
- Motor difficulties
- Small head, also called microcephaly
- Feeding problems
- Vision problems
- Behavioral issues

What causes DYRK1A-related syndrome?

Our genes contain the instructions, or code, that tell our cells how to grow, develop, and work. Every child gets two copies of the DYRK1A gene: one copy from their mother, from the egg, and one copy from their father, from the sperm. In most cases, parents pass on exact copies of the gene to their child. But the process of copying genes is not perfect. A change in the genetic code can lead to physical issues, developmental issues, or both.

Sometimes a random change happens in the sperm or egg. This change to the genetic code is called a 'de novo', or new, change. The child can be the first in the family to have the gene change.



De novo changes can take place in any gene. We all have some de novo changes, most of which don't affect our health. But because DYRK1A plays a key role in development, de novo changes in this gene can have a meaningful effect.

Research shows that DYRK1A-related syndrome is often the result of a de novo change in DYRK1A. Many parents who have had their genes tested do not have the DYRK1A gene change found in their child who has the syndrome. In some cases, DYRK1A-related syndrome happens because the change was passed down from a parent.

Why does my child have a change in the DYRK1A gene?

No parent causes their child's DYRK1A-related syndrome. We know this because no parent has any control over the gene 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 gene 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 DYRK1A-related 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 DYRK1A-related syndrome depends on the genes of both birth parents.

- If neither birth parent has the same gene 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 gene 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 DYRK1A-related 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 gene 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 DYRK1A-related syndrome.
- If one birth parent has the same gene change found in their child who has the syndrome, the symptom-free sibling has a small chance of also having the same gene change. If the symptom-free sibling has the same gene change as their sibling who has the syndrome, the symptom-free sibling's chance of having a child who has DYRK1A-related syndrome is 50 percent.

For a person who has DYRK1A-related syndrome, the risk of having a child who has the syndrome is about 50 percent.

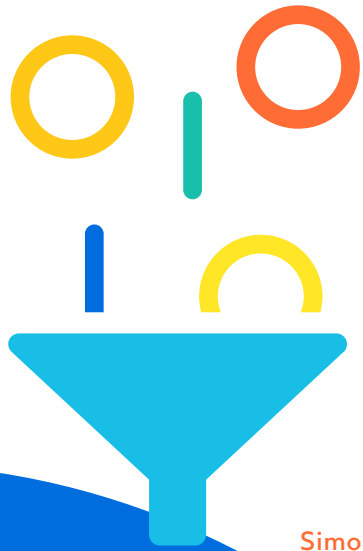
How many people have DYRK1A-related syndrome?

As of 2019, doctors had found about 60 people in the world with changes in the DYRK1A gene. Scientists expect to find more people who have the syndrome as access to genetic testing improves.

Do people who have DYRK1A-related syndrome look different?

People who have DYRK1A-related syndrome may look different. Appearance can vary and can include some but not all of these features:

- Deep-set eyes that look hooded
- Narrow forehead
- Larger than average brow with high hairline
- Tube-shaped nose
- Larger than average nasal bridge
- Lower jaw set back from upper jaw
- Small chin



How is DYRK1A-related syndrome treated?

Scientists and doctors have only just begun to study DYRK1A-related 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
- Development 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 DYRK1A-related 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.

DYRK1A-related syndrome is very rare. Doctors and scientists have just recently begun to study it. As of 2019, studies found around 60 people who have DYRK1A-related 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.

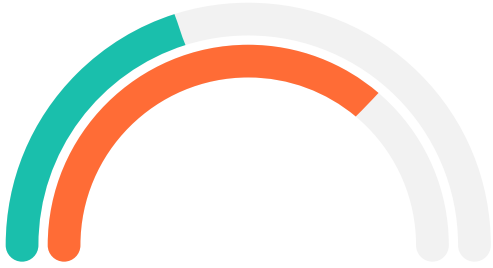
Behavior and development concerns linked to DYRK1A-related syndrome

Speech and Learning

Almost everyone who has DYRK1A-related syndrome has speech delay and intellectual disability.

Behavior

About 40 percent of people who have the condition also have autism. About 70 percent have some autism-like behaviors, such as repetitive movements, limited eye contact, and inappropriate laughter.



40% have autism
70% have autism-like behaviors

Mental health

About one-third of people who have the condition are hyperactive.

Nearly 30 percent show signs of anxiety.

Medical and physical concerns linked to DYRK1A-related syndrome

Brain

Almost everyone who has DYRK1A-related syndrome has a small head. Seventy percent of people who have the condition have seizures.

Motor concerns

Almost everyone who has DYRK1A-related syndrome has motor challenges.

Feeding and digestion issues

More than 90 percent of people who have the condition have feeding issues.

90%

Eyes and eyesight

Eighty percent have vision problems.

80%

Ears and hearing

About one-half of those who have the condition have an unusual shape to their ears, with the outer edge of the ear appearing thick and overfolded.

- **DYRK1A Syndrome International Association**
  www.dyrk1a.org

Sources and References

The content in this guide comes from published studies about DYRK1A-related syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.

- Bronicki LM. *et al. European Journal of Human Genetics*, **23**, 1482-1487, (2015). Ten new cases further delineate the syndromic intellectual disability phenotype caused by mutations in DYRK1A www.ncbi.nlm.nih.gov/pubmed/25920557

- Ji J. *et al. European Journal of Human Genetics*, **23**, 1473-1481, (2015). DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies www.ncbi.nlm.nih.gov/pubmed/25944381

- Earl RK. *et al. Molecular Autism*, **8**, 54, (2017). Clinical phenotype of ASD-associated DYRK1A haploinsufficiency www.ncbi.nlm.nih.gov/pubmed/29034068

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