



CSNK2A1-related 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 CSNK2A1-related syndrome.





What is CSNK2A1-related syndrome?

CSNK2A1-related syndrome happens when there are changes to the CSNK2A1 gene. These changes can keep the gene from working as it should.

Key role

The CSNK2A1 gene plays different roles in the body, including helping to control the birth of new cells and helping to control the day to night cycle of cells.

Symptoms

Because the CSNK2A1 gene is important in the development and function of brain cells, many people who have CSNK2A1-related syndrome have:

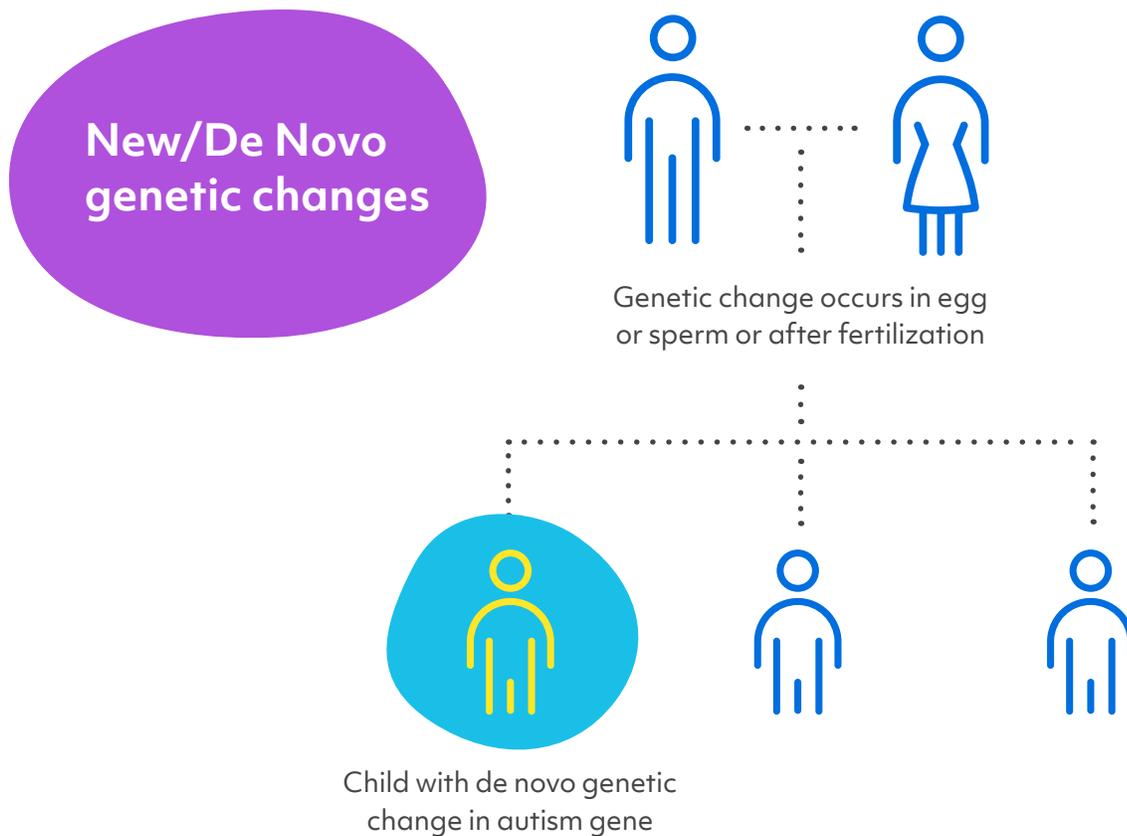
- Physical changes
- Intellectual disability
- Other developmental issues

CSNK2A1-related syndrome is also called Okur-Chung neurodevelopmental syndrome.

What causes CSNK2A1-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 CSNK2A1 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.

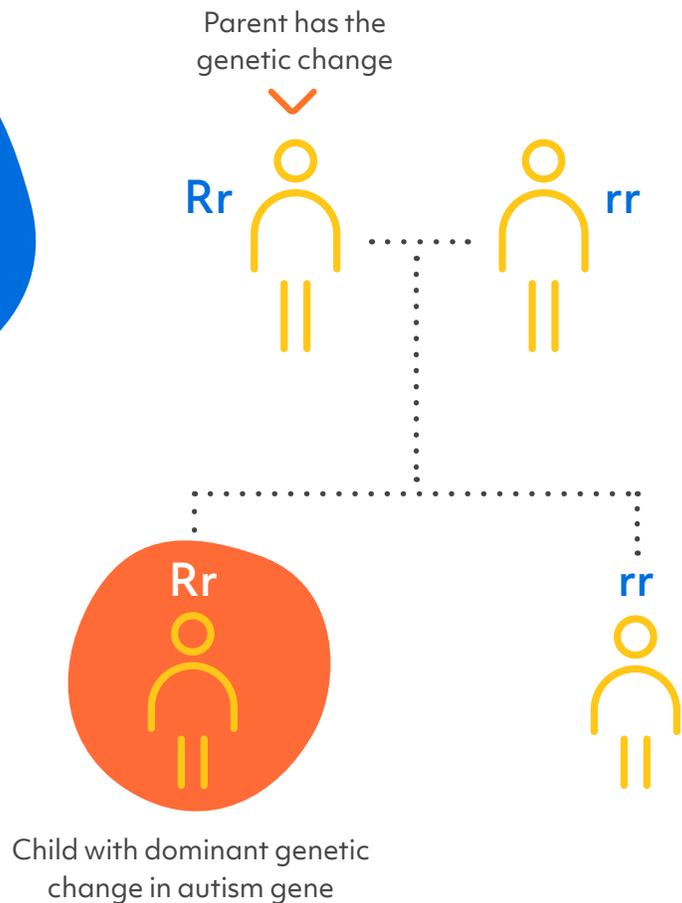


De novo changes can take place in any gene. We all have some de novo changes, most of which don't affect our health. But because CSNK2A1 plays a key role in development, de novo changes in this gene can have a meaningful effect.

Research shows that CSNK2A1-related syndrome is often the result of a de novo change in CSNK2A1. Many parents who have had their genes tested do not have the CSNK2A1 gene change found in their child who has the syndrome. In some cases, CSNK2A1-related syndrome happens because the gene change was passed down from a parent.



Dominant inheritance
Children have a 50% chance of inheriting the genetic change



Why does my child have a change in the CSNK2A1 gene?

No parent causes their child's CSNK2A1-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 CSNK2A1-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 CSNK2A1-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 CSNK2A1-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 CSNK2A1-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 CSNK2A1-related syndrome is 50 percent.

For a person who has CSNK2A1-related syndrome, the risk of having a child who has the syndrome is about 50 percent.

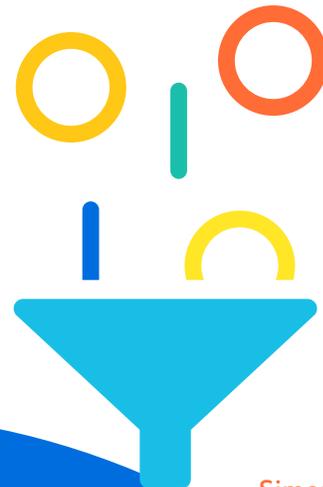
How many people have CSNK2A1-related syndrome?

As of 2019, in the medical research, scientists have described about 27 people in the world with changes in the CSNK2A1 gene. More people have been diagnosed with the syndrome. The first case of CSNK2A1-related syndrome was described in 2016. Scientists expect to find more people who have the syndrome as access to genetic testing improves.

Do people who have CSNK2A1-related syndrome look different?

People who have CSNK2A1-related syndrome may look different. Appearance can vary and can include some but not all of these features:

- Short height
- Smaller than average head size, also called microcephaly
- Round face
- Arched eyebrows
- A fold of skin from the upper eyelid, also called epicanthic folds
- Low set ears and changes in the ear folds
- Wide nasal bridge
- Smaller than average jaw, also called micrognathia



How is CSNK2A1-related syndrome treated?

Scientists and doctors have only just begun to study CSNK2A1-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 CSNK2A1-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](https://www.epilepsy.com/learn/types-seizures).

CSNK2A1-related syndrome is very rare. Doctors and scientists have just recently begun to study it. The information below is based on medical research describing around 27 people who have CSNK2A1-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 CSNK2A1-related syndrome



Almost everyone who has CSNK2A1-related syndrome has **intellectual disability**.

Speech

Speech delay is common in people who have the syndrome.

Sleep

More than one-half have **sleep issues**.

Behavior



7 out of 25 have **Autism**

More than one-quarter have **autism or features of autism**.



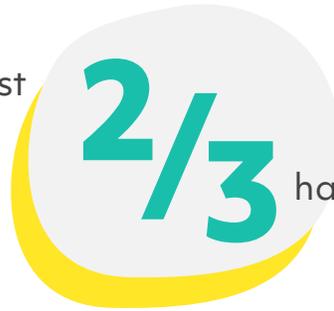
4 out of 25 have **ADHD**

Fewer than 20 percent have **attention deficit hyperactivity disorder**, also called ADHD.

Medical and physical concerns linked to CSNK2A1-related syndrome

Muscle tone

Almost



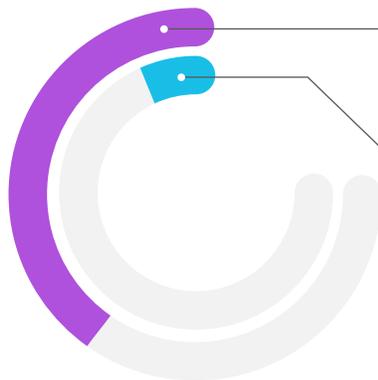
have **low muscle tone**.

Sitting and walking



Some people who have the syndrome are **slow to begin walking**.

Joints and spine



About one-half have some **changes in the skeleton**.

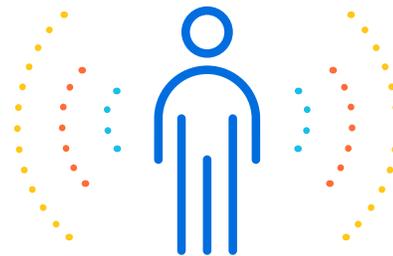
3 out of 26 have a curved spine, also called **scoliosis**.

Medical and physical concerns linked to CSNK2A1-related syndrome



Growth

More than one-third have a **small head size**.



Brain

Fewer than one-quarter have **seizures**.



Feeding and digestion issues

Some people report **issues with feeding and digestion**. This can include acid reflux, constipation, difficulties swallowing, and difficulties controlling the lips, tongue, and jaw muscle, also called **oromotor delay**.



Birth defects

About one-quarter are born with **heart issues**. This can include a hole in the heart, also known as an atrial septal defect.



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

CSNK2A1 Foundation
www.csnk2a1foundation.org



Simons Searchlight is another research program sponsored and run by the Simons Foundation Autism Research Initiative, also known as SFARI. As part of the next step in your research journey, Simons Searchlight offers you the opportunity to partner with scientists and other families who have the same gene change.

Simons Searchlight has made a **Facebook page** for people who have the CSNK2A1 gene change that you and your family may use to connect with other CSNK2A1 families as they are identified:

www.facebook.com/groups/searchlight.CSNK2A1



Simons Searchlight page on CSNK2A1
www.simonssearchlight.org/research/what-we-study/csnk2a1

Sources and References

The content in this guide comes from published studies about CSNK2A1-related syndrome. Below you can find details about each study, as well as links to summaries.

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