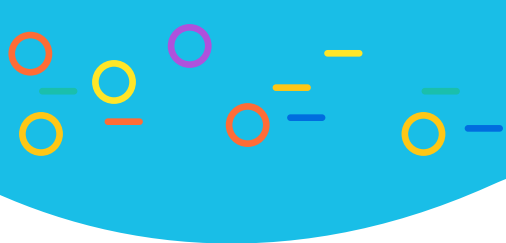




ARID1B-related syndrome





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 ARID1B-related syndrome.





What is ARID1B-related syndrome?

ARID1B-related syndrome happens when there are changes to the ARID1B gene. These changes can keep the gene from working as it should.

Key role

The ARID1B gene helps to control other genes that are important for brain growth.

Symptoms

Because the ARID1B gene is important in the development and function of brain cells, many people who have ARID1B-related syndrome have:

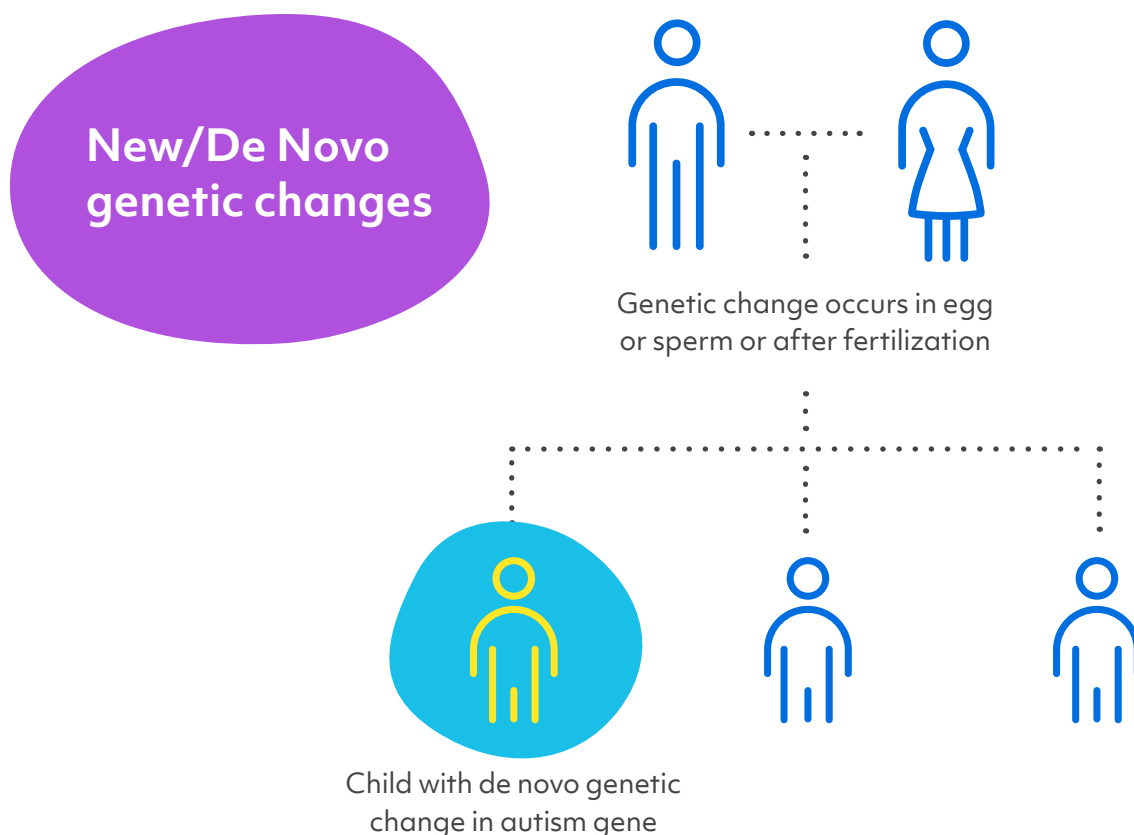
- Intellectual disability
- Behavior issues, including autism and attention deficit hyperactivity disorder, also known as ADHD
- Seizures
- Feeding difficulties
- Hearing issues

Symptoms can range from mild to severe. People who have severe symptoms often have a condition called Coffin-Siris syndrome. Please see the Coffin-Siris syndrome guide for more information. People who have mild symptoms have ARID1B-related syndrome, which is described in this guide. ARID1B-related syndrome is tied to intellectual disability and may or may not include changes in physical appearance.

What causes ARID1B-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 ARID1B 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.



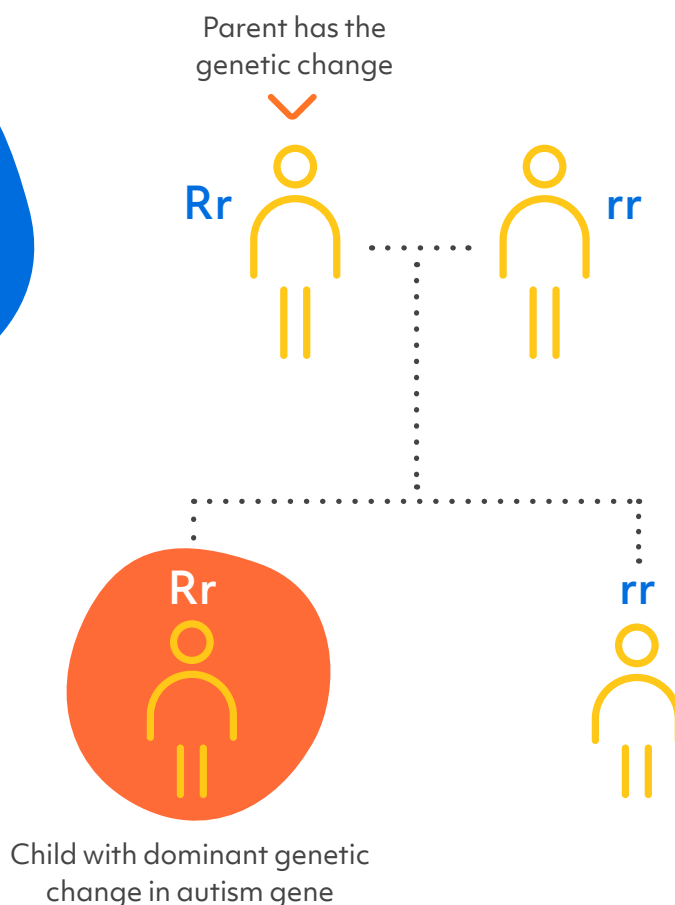
De novo changes can take place in any gene. We all have some de novo changes, most of which don't affect our health. But because ARID1B plays a key role in development, de novo changes in this gene can have a meaningful effect.

Research shows that ARID1B-related syndrome is often the result of a de novo change in ARID1B. Many parents who have had their genes tested do not have the ARID1B gene change found in their child who has the syndrome. In some cases, ARID1B-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 ARID1B gene?

No parent causes their child's ARID1B-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 ARID1B-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 ARID1B-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 ARID1B-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 ARID1B-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 ARID1B-related syndrome is 50 percent.

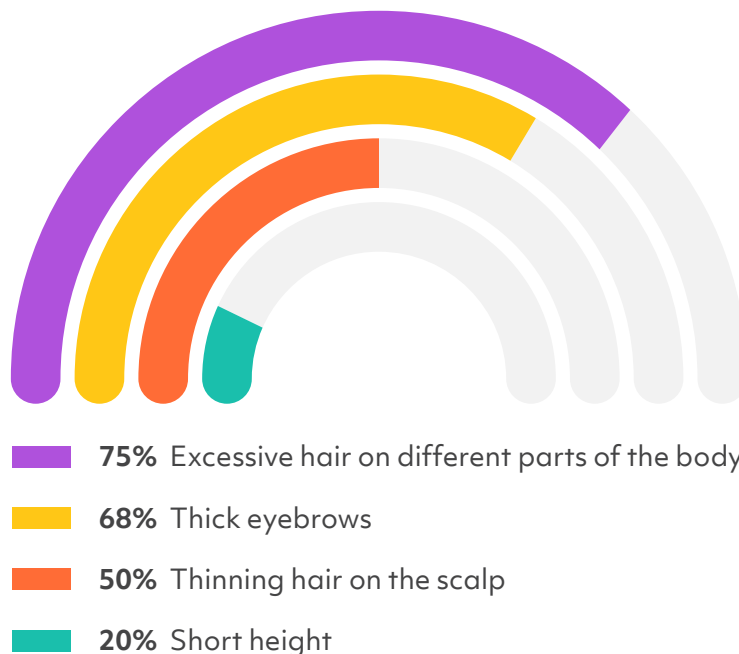
For a person who has ARID1B-related syndrome, the risk of having a child who has the syndrome is about 50 percent.

How many people have ARID1B-related syndrome?

As of 2019, about 65 people in the world with changes in the ARID1B gene had been described in the medical literature. The first case of ARID1B-related syndrome was described in 2012. Scientists expect to find more people who have the syndrome as access to genetic testing improves.

Do people who have ARID1B-related syndrome look different?

People who have ARID1B-related syndrome may look different. Appearance can vary and can include some but not all of these features:





How is ARID1B-related syndrome treated?

Scientists and doctors have only just begun to study ARID1B-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 ARID1B-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: www.epilepsy.com/learn/types-seizures.

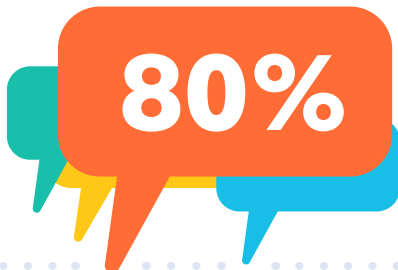
ARID1B-related syndrome is very rare. Doctors and scientists have just recently begun to study it. As of 2019, studies described around 65 people who have ARID1B-related syndrome.

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 ARID1B-related syndrome

Learning

Everyone who has ARID1B-related syndrome has some degree of **intellectual disability**.

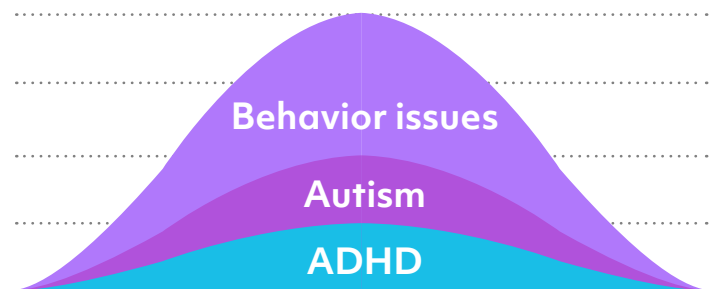


Speech

About 80 percent have **speech delay**.

Behavior

About 80 percent have **behavior issues**. Of those, about 50 percent have **autism** and 25 percent have **ADHD**.



About 15 percent have **sleep apnea**.

Medical and physical concerns linked to ARID1B-related syndrome



Sitting and walking

Almost everyone who has ARID1B-related syndrome has **motor delays**.

Muscle tone

About 80 percent have **low muscle tone**.



**About
75%**

Feeding and digestion issues

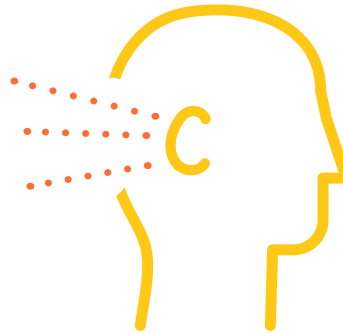
About 75 percent of young infants who have ARID1B-related syndrome have **feeding difficulties**.

Medical and physical concerns linked to ARID1B-related syndrome



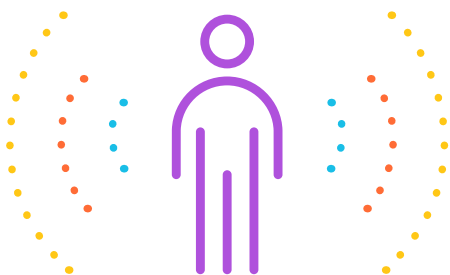
Eyes and eyesight

About 50 percent have vision problems.



Ears and hearing

About 25 percent have hearing loss.



Brain

About 25 percent have **seizures**.



Growth

About 20 percent are **short**.



Where can I find support and resources?

At this time, we are not aware of support groups for families with changes in the ARID1B gene. More families who have gene changes in ARID1B will likely be found in the future.



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 is a registry for more than 150 genetic changes that are associated with neurodevelopmental conditions, including autism spectrum disorder. Simons Searchlight makes it easier for researchers to access the information they need to advance research on a condition.

To register for Simons Searchlight, go to the Simons Searchlight website at www.simonssearchlight.org and click “Join Us Today”.

- **Learn more about Simons Searchlight**
www.simonssearchlight.org/frequently-asked-questions
- **Simons Searchlight webpage with more information on ARID1B**
www.simonssearchlight.org/research/what-we-study/arid1b
- **Simons Searchlight ARID1B Facebook community**
www.facebook.com/groups/searchlight.arid1b

Sources and References

The content in this guide comes from published studies about ARID1B-related syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.

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www.ncbi.nlm.nih.gov/pmc/articles/PMC6752273
- Vergano SA. *et al. Gene Reviews*, (2019). ARID1B-related disorder
www.ncbi.nlm.nih.gov/books/NBK541502



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