



## DSCAM-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 2021. 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 this gene change.





## What is DSCAM-related syndrome?

You are receiving this information because you or your child has a change in a newly discovered autism gene, DSCAM.

Changes in the DSCAM gene are linked to intellectual disability and autism. Because DSCAM is so new in autism, we don't have specific information on what medical issues you might expect now and in the near future. We can tell you that DSCAM plays an important role in brain development and is associated with autism. The gene is also likely associated with language and learning.

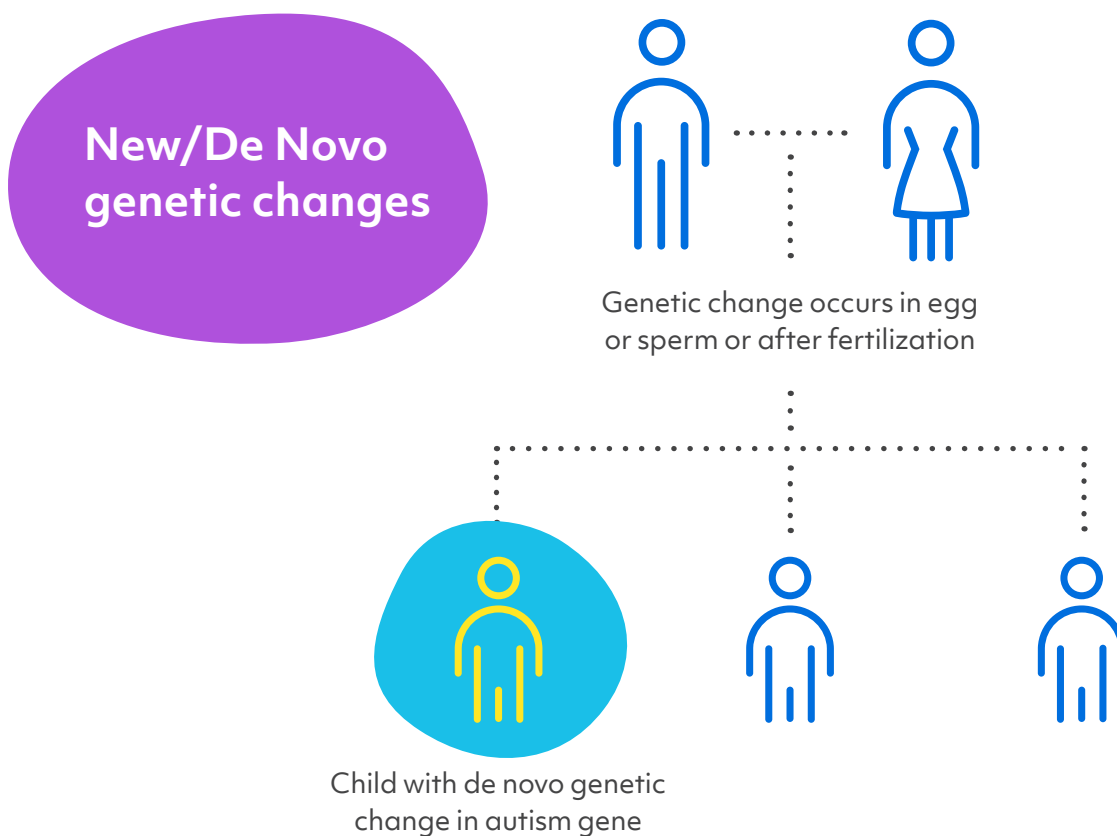
A medical article describing one person with a change in the DSCAM gene shows that this person had autism, mild developmental delay, repetitive behavior, hyperactive behavior, attention issues, and obsessive behavior.

You, or your family member, are one of what may be a very small number of people in the world with autism who have a change in the DSCAM gene. Scientists expect to find more people who have changes in DSCAM as access to genetic testing improves.

## What causes DSCAM-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 DSCAM 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.



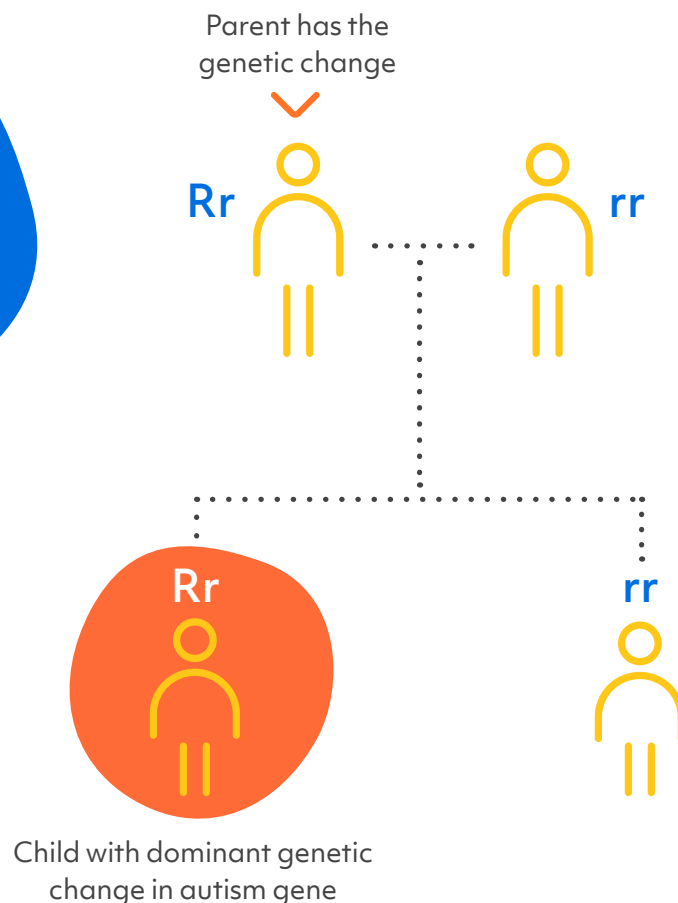
New changes can take place in any gene. We all have some new changes, most of which don't affect our health. But because DSCAM plays a key role in brain development, new changes in this gene can have a meaningful effect.

Research shows that changes in DSCAM are often the result of a new change in the gene. Many parents who have had their genes tested do not have the DSCAM gene change found in their child who has the syndrome. In some cases, changes in DSCAM happen because the gene change was passed down from a parent. This is called dominant inheritance.



## Dominant inheritance

Children have a 50% chance of inheriting the genetic change



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## Why does my child or I have a change in the DSCAM gene?

No parent causes their child's changes in DSCAM. 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.





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## What are the chances that other family members or future children will have changes in DSCAM?

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 changes in DSCAM 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 changes in DSCAM, 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 changes in DSCAM.
- 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 changes in DSCAM is 50 percent.

For a person who has changes in DSCAM, the risk of having a child who has the syndrome is about 50 percent.

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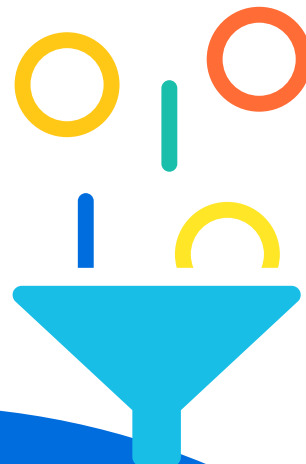
## How many people have a de novo gene change in DSCAM?

As of 2021, researchers had described fewer than 10 people in the world with autism or neurodevelopmental disorders and changes in the DSCAM gene. The first case of this condition was described in 2014. Scientists expect to find more people who have the syndrome as access to genetic testing improves.

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## Do people who have de novo changes in the DSCAM gene look different?

We do not yet know if people who have de novo changes in the DSCAM gene look consistently different from others.







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## How are people who have changes in DSCAM treated?

Scientists and doctors have only just begun to study people who have changes in the DSCAM gene. At this point, there are no medicines designed to treat the condition. 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 people who have autism begin as early as possible, ideally before a child begins school.

If seizures happen, consult a neurologist. There are many different 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](http://www.epilepsy.com/learn/types-seizures).

A doctor may also refer people to other specialists as needed.



## Where can I find support and resources?

**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](http://www.simonssearchlight.org) and click “Join Us Today”.

- Learn more about Simons Searchlight  
[www.simonssearchlight.org/frequently-asked-questions](http://www.simonssearchlight.org/frequently-asked-questions)
- Simons Searchlight webpage with more information on DSCAM  
[www.simonssearchlight.org/research/what-we-study/dscam](http://www.simonssearchlight.org/research/what-we-study/dscam)
- Simons Searchlight DSCAM Facebook community  
[www.facebook.com/groups/649966181781643](https://www.facebook.com/groups/649966181781643)

## Sources and References

The information in this guide comes from a published medical article about people who have autism and who have de novo genetic changes in DSCAM. Below you can find details about the article, as well as a link to the full article.

- Wang T. *et al.* *Nature Communications*, **7**, 13316, (2016). De novo genic mutations among a Chinese autism spectrum disorder cohort, [www.nature.com/articles/ncomms13316](http://www.nature.com/articles/ncomms13316)



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