



# SHANK2-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 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 SHANK2-related syndrome.





## What is SHANK2-related syndrome?

SHANK2-related syndrome happens when there are changes to the SHANK2 gene. These changes can keep the gene from working as it should.

### Key role

The SHANK2 gene plays a key role in communication between brain cells. SHANK2 is closely related to SHANK3, another well-known autism risk gene. There are hundreds of autism risk genes and none of them account for a large number of cases. But SHANK3 is one of the more common autism risk genes, with some studies estimating that up to 1% of people who have autism have changes in SHANK3.

### Symptoms

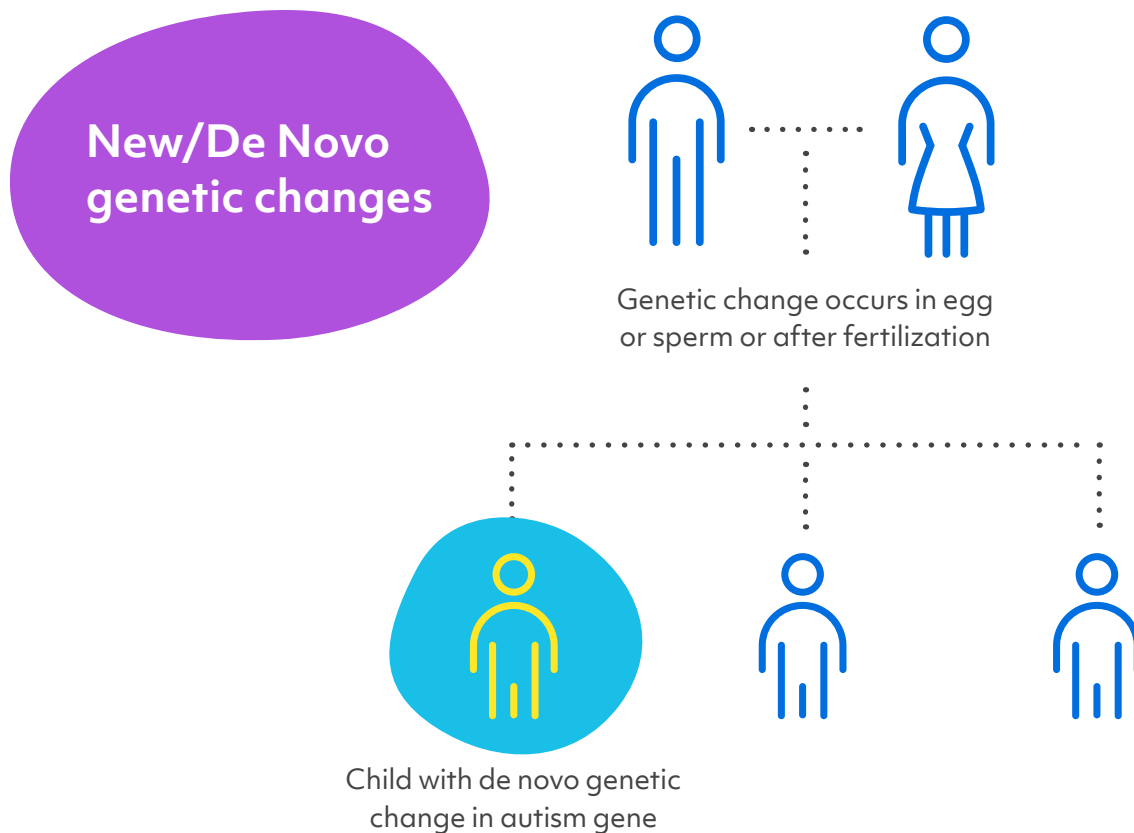
Because the SHANK2 gene is important in the development and function of brain cells, many people who have SHANK2-related syndrome have:

- Intellectual disability
- Autism

## What causes SHANK2-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 SHANK2 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.

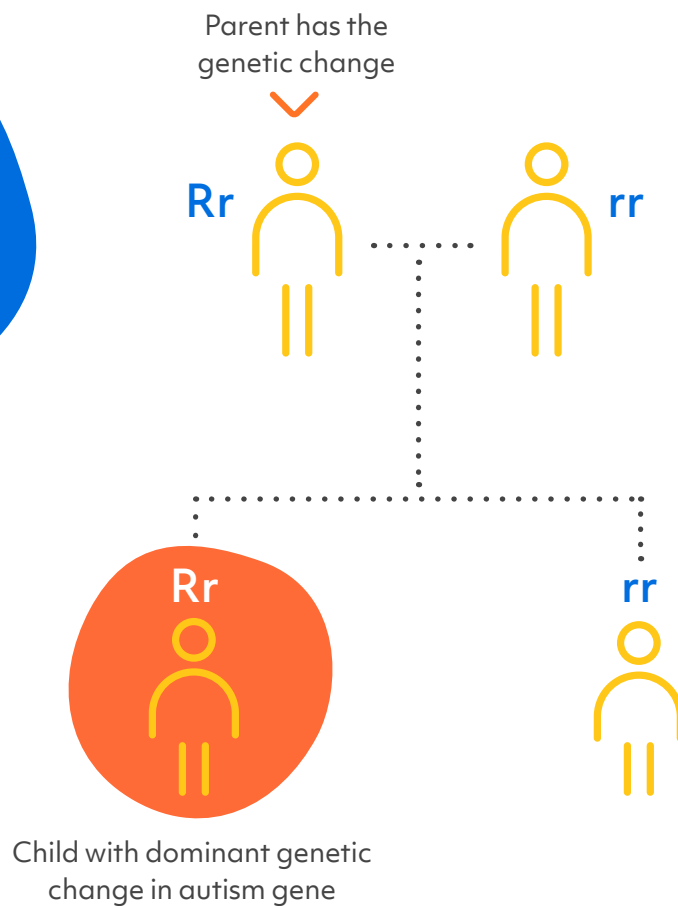


De novo changes can take place in any gene. We all have some de novo changes, most of which don't affect our health. But because SHANK2 plays a key role in development, de novo changes in this gene can have a meaningful effect.

Research shows that SHANK2-related syndrome is often the result of a de novo change in SHANK2. Many parents who have had their genes tested do not have the SHANK2 gene change found in their child who has the syndrome. In some cases, SHANK2-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




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## Why does my child or I have a change in the SHANK2 gene?

No parent causes their child's SHANK2-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.





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## What are the chances that other family members or future children will have SHANK2-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 SHANK2-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 SHANK2-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 SHANK2-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 SHANK2-related syndrome is 50 percent.

For a person who has SHANK2-related syndrome, the risk of having a child who has the syndrome is about 50 percent.

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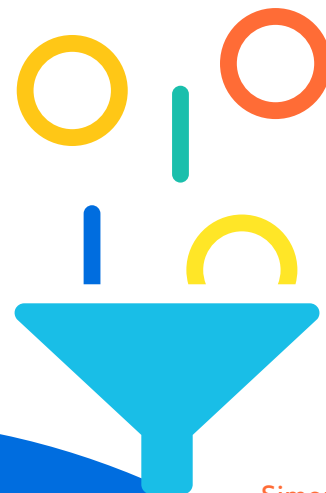
## How many people have SHANK2-related syndrome?

As of 2020, doctors had described about 10 people in the world with changes in the SHANK2 gene. The first case of SHANK2-related syndrome was described in 2010. Scientists expect to find more people who have the syndrome as access to genetic testing improves.

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## Do people who have SHANK2-related syndrome look different?

People who have SHANK2-related syndrome do not look very different.







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## How is SHANK2-related syndrome treated?

Scientists and doctors have only just begun to study SHANK2-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 SHANK2-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).

SHANK2-related syndrome is very rare. Doctors and scientists have just recently begun to study it. As of 2020, doctors have described about 10 people who have SHANK2-related syndrome.

This section includes a summary of information from a major published article. It highlights how many people have different symptoms. To learn more about the article, see the [Sources and references](#) section of this guide.

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## Behavior and development concerns linked to SHANK2-related syndrome



### Behavior

A study of 10 people who have SHANK2-related syndrome found that 7 of them have **autism**.

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### Learning

A study of 10 people who have SHANK2-related syndrome found that 4 of them have **intellectual disability**.





## Where can I find support and resources?

### **Shank2 Gene Mutation Facebook Page**

[www.facebook.com/theshank2foundation](http://www.facebook.com/theshank2foundation)



**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 SHANK2**  
[www.simonssearchlight.org/research/what-we-study/shank2](http://www.simonssearchlight.org/research/what-we-study/shank2)
- **Simons Searchlight SHANK2 Facebook community**  
[www.facebook.com/groups/857070631358977](http://www.facebook.com/groups/857070631358977)

## Sources and References

The content in this guide comes from a published study about SHANK2-related syndrome. Below you can find details about the study, as well as a link to a summary.

- Berkel S. *et al. Nature Genetics*, **42**, 489-491, (2010). Mutations in the SHANK2 synaptic scaffolding gene in autism spectrum disorder and mental retardation  
[www.ncbi.nlm.nih.gov/20473310-mutations-in-the-shank2-synaptic-scaffolding-gene-in-autism-spectrum-disorder-and-mental-retardation](http://www.ncbi.nlm.nih.gov/20473310-mutations-in-the-shank2-synaptic-scaffolding-gene-in-autism-spectrum-disorder-and-mental-retardation)



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