GIGYF1-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 the genetic result.
What is my genetic result?

You are receiving this information because you or your child has a gene change in a newly discovered autism gene, GIGYF1.

Your or your child’s gene change is ‘de novo’, or new. That is, it was not inherited from a parent.

Because GIGYF1 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 GIGYF1 plays an important role in brain development and is associated with autism. The gene is also likely associated with language and learning.

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 gene change in GIGYF1. Scientists expect to find more people who have changes in GIGYF1 as access to genetic testing improves.
What causes de novo gene changes?

Our genes contain the instructions, or code, that tell our cells how to grow, develop, and work. Every child gets two copies of the GIGYF1 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/De Novo genetic changes

Genetic change occurs in egg or sperm or after fertilization

Child with de novo genetic change in autism gene
New changes can take place in any gene. We all have some new changes, most of which don’t affect our health. But because the GIGYF1 gene plays a key role in development, new changes in this gene can have a meaningful effect.

Research shows that changes in GIGYF1 is often the result of a new change in GIGYF1. Many parents who have had their genes tested do not have the GIGYF1 gene change found in their child who has the syndrome. In some cases, changes in GIGYF1 happen because the gene change was passed down from a parent. This is called dominant inheritance.
Why does my child or I have a change in the GIGYF1 gene?

No parent causes their child’s de novo gene change. 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 a de novo gene change in the GIGYF1 gene?

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 a gene change in GIGYF1 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 gene change 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 gene change is 50 percent.

For a symptom-free sibling, a brother or sister, of someone who has a gene change in GIGYF1, the risk of having a child who has a similar gene change depends on the symptom-free sibling’s genes and their parents’ genes.

- If neither parent has the same change to GIGYF1 found in their child who has the gene change, the symptom-free sibling has a nearly 0 percent chance of having a child who has the gene change.

- If one birth parent has the same gene change found in their child who has the gene change, 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 gene change, the symptom-free sibling’s chance of having a child who has a similar gene change is 50 percent.

For a person who has a gene change in GIGYF1, the risk of having a child who has a similar gene change is about 50 percent.
How many people have a de novo gene change in the GIGYF1 gene?

As of 2019, fewer than 12 people in the world with changes in GIGYF1 gene and autism or neurodevelopmental disorders have been described in medical research. The first case of this condition was described in 2014. Scientists expect to find more people who have the condition as access to genetic testing improves.

Do people who have de novo changes in the GIGYF1 gene look different?

We do not yet know if people who have de novo changes in the GIGYF1 gene look consistently different from others.
How are people who have changes in GIGYF1 treated?

Scientists and doctors have only just begun to study people who have changes in the GIGYF1 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 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.

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 and click “Join Us Today”.

- Learn more about Simons Searchlight
  www.simonssearchlight.org/frequently-asked-questions

- Simons Searchlight webpage with more information on GIGYF1
  www.simonssearchlight.org/research/what-we-study/gigyf1

- Simons Searchlight Facebook group
  www.facebook.com/groups/447939852469636
Sources and References

The information in this guide comes from published studies about people who have autism who have de novo gene changes in GIGYF1. Below you can find details about each study, as well as links to summaries, or in some cases the full article.

  www.ncbi.nlm.nih.gov/pubmed/25363768

  www.ncbi.nlm.nih.gov/pubmed/25961944