EIF3F-related syndrome

SPARK
Simons Powering Autism Research

SIMONS SEARCHLIGHT
Driven by science. United by hope.

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 EIF3F-related syndrome.

What is EIF3F-related syndrome?

EIF3F-related syndrome happens when there are changes to the EIF3F gene. These changes can keep the gene from working as it should. This guide describes the symptoms that are linked to a specific change in the EIF3F gene.

Key role

The EIF3F gene helps to make proteins, which carry out many important jobs in the cell.

Symptoms

Because the EIF3F gene is important in the development and function of brain cells, many people who have EIF3F-related syndrome have:

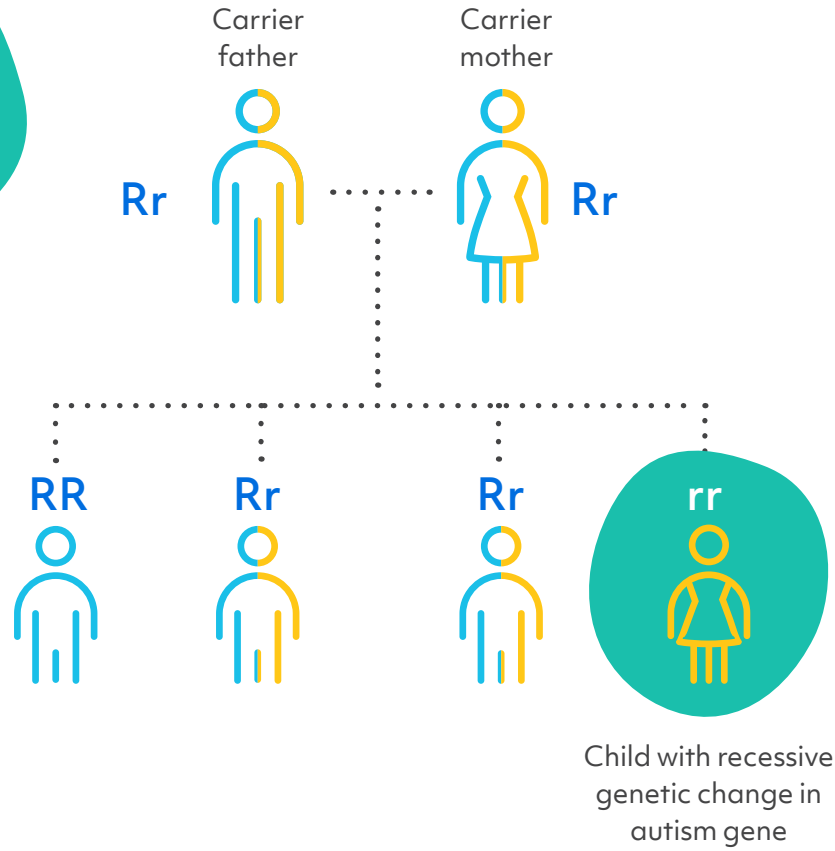
- Intellectual disability
- Seizures

What causes EIF3F-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 EIF3F gene: one copy from their mother, from the egg, and one copy from their father, from the sperm.

Some people have changes to the gene that prevent it from working properly. A change in one copy of the EIF3F gene has little or no effect on their health — one working copy is enough. People who have one working copy of the gene and one non-working copy of the gene are called 'carriers'. Some people have genes where both copies do not work as they should. In these cases, the person has inherited non-working copies of the gene from both parents. This can lead to physical issues, developmental issues, or both.

Recessive genetic changes



Child with recessive genetic change in autism gene

Why does my child or I have a change in the EIF3F gene?

No parent causes their child's EIF3F-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.

What are the chances that other family members or future children will have EIF3F-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 EIF3F-related syndrome is almost always 25 percent, but it also depends on the genes of both birth parents.

- The chance of two carrier parents having a child who is also a carrier but does not have the syndrome is 50 percent. The chance of them having a child who is not a carrier and does not have the syndrome is 25 percent.

For a person who has EIF3F-related syndrome, the risk of having a child who has the syndrome depends on their partner.

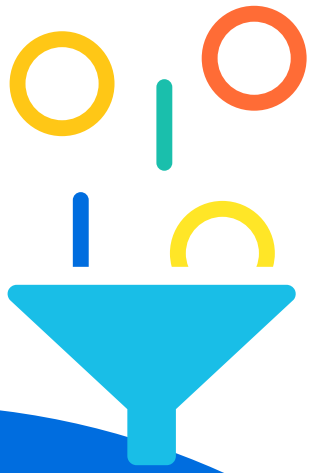
- If their partner is a carrier, they have a 50 percent chance of having a child who has the syndrome and a 50 percent chance of having a child who is a carrier.
- If their partner is not a carrier, they have a 0 percent chance of having a child who has the syndrome and a 100 percent chance of having a child who is a carrier.

How many people have EIF3F-related syndrome?

As of 2020, about 9 people in the world with changes in the EIF3F gene had been described in medical research. Scientists expect to find more people who have the syndrome as access to genetic testing improves.

Do people who have EIF3F-related syndrome look different?

People who have EIF3F-related syndrome do not look very different.



How is EIF3F-related syndrome treated?

Scientists and doctors have only just begun to study EIF3F-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 EIF3F-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.

EIF3F-related syndrome is very rare. Doctors and scientists have just recently begun to study it. As of 2020, studies found around 9 people who have this change in the EIF3F gene.

This section includes a summary of information from a major article published in 2018. It highlights how many people have different symptoms. To learn more about the article, see the Sources and references section of this guide.

Behavior and development concerns linked to EIF3F-related syndrome

Learning

had **intellectual disability**

Behavior

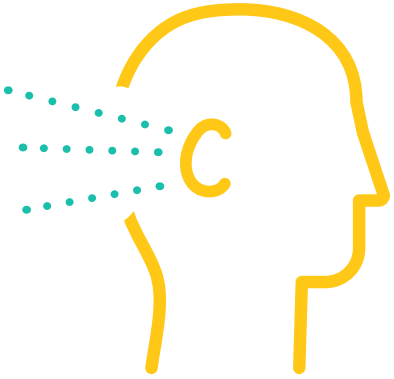
had **behavior challenges**

Medical and physical concerns linked to EIF3F-related syndrome

Brain

6 out of **9** had **seizures**

Ears and hearing



3 out of **9** had **hearing loss**

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 200 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 EIF3F**
  www.simonssearchlight.org/research/what-we-study/eif3f
- **Simons Searchlight EIF3F Facebook community**
  www.facebook.com/groups/268676524180871

Sources and References

The content in this guide comes from a published study about EIF3F-related syndrome. Below you can find details about the study, as well as a link to the full article.

- Martin HC. *et al. Science*, **362**, 1161-1164, (2018). Quantifying the contribution of recessive coding variation to developmental disorders
  www.science.org/doi/10.1126/science.aar6731

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