SCN1A-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 SCN1A-related syndrome.
What is SCN1A-related syndrome?

SCN1A-related syndrome happens when there are changes to the SCN1A gene. These changes can keep the gene from working as it should.

Key role
The SCN1A gene produces a protein that sits in the surface of brain cells and allows sodium to enter the cell. This protein is important for brain cells to make and transmit signals between cells. The protein is essential for these brain cells to work properly.

Symptoms
Because the SCN1A gene is important in the development and function of brain cells, many people who have SCN1A-related syndrome have:

- Epilepsy
- Developmental delay and/or intellectual disability
- Autism spectrum disorder or autistic features
- Movement problems
- Concerns with sleep
People who have gene changes in SCN1A may have different types of seizure disorders:

1. **Dravet syndrome**: In this condition, severe seizures begin early in life. People who have this condition often have cognitive impairment. Research studies suggest that 33 to 90 percent of people who have changes in SCN1A have Dravet syndrome.

2. **Generalized epilepsy with febrile seizures plus (GEFS+)**: Symptoms of this condition vary from person to person. Some people have seizures with fever. Other people have more severe epilepsy. Studies show that this condition affects 5 to 10 percent of people who have changes in SCN1A.

Other types of seizure disorders that affect people who have changes in SCN1A include: myoclonic astatic epilepsy (MAE), Lennox-Gastaut syndrome, infantile spasms, and epilepsy with focal seizures. Different family members who have changes in SCN1A can have different symptoms.
What causes SCN1A-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 SCN1A 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 genetic 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
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 SCN1A plays a key in development, de novo changes in this gene can have a meaningful effect.

Research shows that SCN1A-related syndrome is often the result of a de novo change in SCN1A. Many parents who have had their genes tested do not have the SCN1A gene change found in their child who has the syndrome. In some cases, SCN1A-related syndrome happens because the gene change was passed down from a parent.
Why does my child have a change in the SCN1A gene?

No parent causes their child’s SCN1A-related syndrome. We know this because no parent has any control over the genetic 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 genetic 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 SCN1A-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 SCN1A-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 SCN1A-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 SCN1A-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 SCN1A-related syndrome is 50 percent.

For a person who has SCN1A-related syndrome, the risk of having a child who has the syndrome is about 50 percent.
How many people have SCN1A-related syndrome?

SCN1A-related syndrome occurs in 1 in 20,900 births. The first case of SCN1A-related syndrome was described in 2000. Scientists expect to find more people who have the syndrome as access to genetic testing improves.

Do people who have SCN1A-related syndrome look different?

People who have SCN1A-related syndrome generally don’t look physically different.
How is SCN1A-related syndrome treated?

Epilepsy is common with SCN1A-related syndrome. People who have the condition should be under the care of a physician, such as a pediatric epileptologist, who is familiar with the best medications for this disorder. Seizure control is important to prevent severe injuries. However, in some cases, seizures linked to SCN1A-related conditions cannot be completely controlled. Antiepileptic drugs (AEDs) that bind to the GABA receptor may be beneficial. AEDs include clobazam and stiripentol. Levetiracetam is often effective but may make seizures worse in some individuals. Phenobarbital is effective but in some cases is poorly tolerated because of its effects on mental function. A ketogenic diet that is high in fat and low in carbohydrates can decrease how often seizures happen in some people who have the condition. Parents are advised to take a CPR course.

Several anti-epilepsy medications, such as carbamazepine, lamotrigine, and vigabatrin, should be avoided because they can cause or increase myoclonic seizures. Phenytoin should be avoided because it can cause involuntary movements, or choreoathetosis. Rufinamide should be avoided because it may worsen seizures. Acetaminophen should be avoided because it can damage the liver. Activities in which a sudden loss of consciousness could lead to injury or death, such as bathing, swimming, driving, or working or playing at heights should also be avoided. Sleep deprivation can worsen seizures.

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.

It is important to seek care and therapies to help the person's brain and nervous system develop and operate as well as it can. Soon after diagnosis, this can include:

- Brain studies. An electroencephalogram (EEG) may show the overall electrical disturbances and may help a person's doctor learn how to best treat their epilepsy.
How is SCN1A-related syndrome treated?

Right now, there is no specific advice about how to treat other symptoms. 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:

- Genetics consults.
- Development and behavior studies.
- Gastrointestinal issues. Many children who have SCN1A-related syndrome have constipation, gastroesophageal reflux or diarrhea.
- 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 SCN1A-related syndrome should begin as early as possible, ideally before a child begins school.
SCN1A-related syndrome is rare. As of 2019, researchers had found more than 200 people who have the syndrome.

This section includes a summary of information from 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.

### Medical, behavior, and development concerns linked to SCN1A-related syndrome

#### Brain

Almost everyone who has SCN1A-related syndrome has seizures. The seizures fall into different categories:

- **Dravet syndrome:**
  - 33 to 90 percent
  - Seizures begin early in life

- **Generalized epilepsy with febrile seizures (GEFS+):**
  - 5 to 10 percent

- **Febrile seizures:** frequency unknown

- In a study of 164 people who have SCN1A-related disorder:
  - 70% had Dravet syndrome
  - 30% had GEFS+ or febrile seizures

#### Behavior and development

Almost half of people who have Dravet syndrome have behavior problems. This is less common in people who have SCN1A-related disorder but do not have Dravet syndrome.
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 SCN1A
  www.simonssearchlight.org/research/what-we-study/scn1a

- Simons Searchlight Facebook group
  www.facebook.com/groups/468546337324008
Sources and References

The content in this guide comes from published studies about SCN1A-related syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.
