FACTS ABOUT HEALTH CONDITIONS

CAUSED BY CHANGES IN THE *SCN2A* GENE

This fact sheet contains information about the possible impact of a change (variant) in the *SCN2A* gene on your child and family. You can talk about the information in this fact sheet with your paediatrician or GP (family doctor). The links in the fact sheet may help you move forward with family life beyond receiving this rare diagnosis.

This fact sheet relates to health conditions that are due to small variants in the genetic code of the *SCN2A* gene. These changes were identified by a genomic (DNA) test.

Key points

- A change in the *SCN2A* gene can cause a number of different conditions, which may be mild or more severe
- Symptoms of *SCN2A*-related conditions include epilepsy (seizures), developmental delay/intellectual disability, autism spectrum disorders, and movement disorders
- Changes in the *SCN2A* gene that cause milder health problems may be passed on from a less affected parent
- More commonly, the changes are new (*'de novo'*). In this case, it is unlikely that future children will be affected. Genetic counselling before any further pregnancies is recommended
- There are not yet any specific treatments, however, supportive management is available
- Anti-seizure medications can help some children with *SCN2A*-related epilepsy

When a rare condition has been diagnosed

For some families, receiving a genetic diagnosis is a relief. Others may feel overwhelmed and sad. It is very common to have a mixture of thoughts and feelings about the news, and your hopes and expectations for the future may shift and change over time.

While experiences may be shared, individuals and families can respond in different ways and have different information and support needs. Many parents describe an ongoing process of adjusting to a different focus and finding ways to celebrate their child's gains made in their own way and time. It is very important to remember that the diagnosis is only one of many things that make your child unique.

Version 1. June, 2022
SHPN: (HETI) 240998

About the *SCN2A* gene

Genes contain instructions that tell our body how to grow, develop and function. *SCN2A* is a **gene** that tells our body to produce an important sodium channel in the brain. This channel controls the way brain cells talk to each other. Changes in the *SCN2A* gene can cause brain cells to be more or less active than they should be. This in turn can cause epileptic seizures and/or differences in how a child learns and develops.

The *SCN2A* gene is found on chromosome 2. Usually *SCN2A*-related conditions are caused by a single spelling variation in the gene, which means the message is not read or received properly. In some individuals, a section of the chromosome that includes the *SCN2A* gene is missing. This is called a deletion.

In most individuals with *SCN2A*-related conditions, the gene change occurred when the baby was conceived (a new or *'de novo'* variant), and is not seen in the parents. In some of the milder forms, the variant is passed to the child from a parent.

SCN2A-related conditions are **genetic conditions**. This means that the condition was not caused by anything the mother or father did before the baby was conceived, during pregnancy or birth, or after the baby was born. *SCN2A*-related conditions are rare, affecting 1 in 78,000 children, (although this is likely an underestimate).

What could a change in the *SCN2A* gene mean for my child?

A change in the *SCN2A* gene can affect children in different ways. Some are more severely affected than others. There may be a range of signs and symptoms even in children with the same genetic variant.

Changes in the *SCN2A* gene can cause a number of conditions that affect the way the brain works. These are also known as neurodevelopmental conditions. As the neurological system affects the whole body, the symptoms of *SCN2A* may be seen across the entire body.

Many children with *SCN2A*-related conditions are slower to reach their physical milestones (sitting, crawling, walking, running), and have problems with learning. This can range from specific learning difficulties to an intellectual disability, which range from mild to severe.

Children may have problems with their movement, such as uncontrolled muscle tightening (dystonia), irregular and unpredictable muscle movements (chorea) or slurred speech and uncoordinated movement (ataxia).

Behaviour is also affected in many children. Some children with *SCN2A*-related conditions are diagnosed with autism, or have some autism-like behaviours. These symptoms generally start in early childhood.

Many children with changes in the *SCN2A* gene have seizures (epilepsy). Whether or not a child has seizures, the types of seizures and whether medication can control those seizures will determine how their condition is managed.

SCN2A-related conditions with epilepsy

Children with epilepsy may have seizures that range from self-limiting (stop without treatment) to severe and difficult to control with medication. These seizures may start shortly after the child is born or as late as 17 years of age.

Seizure type can vary – in some children the seizures may affect certain parts of the body only (focal epilepsy), while in other children the seizure will affect the whole body (generalised epilepsy). The seizures may be difficult to control and treat, and some medications can make seizures worse. It is advisable for the paediatrician to seek the opinion of a paediatric neurologist through the usual referral pathways.

SCN2A-related conditions without epilepsy

Many children with changes in the *SCN2A* gene do not have epilepsy. However, they may develop other symptoms, including reflux and constipation, uncontrolled muscle tightening (dystonia) and periods of unsteadiness (episodic ataxia). These episodes, which may last minutes to hours, usually start occurring between the ages of 10 months and 14 years.

Version 1. June, 2022.

Caring for a child with an *SCN2A*-related condition

At this point in time, it is not possible to reverse or directly repair this gene change. It is also not possible to accurately predict the level of care your child will require through to adulthood. Your child's individual needs and strengths will become more obvious over time, which will help with planning for the future.

For children with epilepsy or movement disorders, a visit to a paediatric neurologist is recommended.

Your child's development may be helped through early use of therapy services such as physiotherapy, and treating symptoms if/when they arise. It is likely that many different health professionals will be involved in caring for your child. Your paediatrician or GP will arrange referrals to other health professionals as needed and help with applications for service funding through the National Disability Insurance Scheme (NDIS).

Good communication with the health professionals caring for your child is important to establish common goals, trust and shared responsibility. We encourage you to ask questions and express your concerns as the expert carer for your child.

Management recommendations

As many health or developmental problems are not immediately obvious, your child will need to be checked by their paediatrician at diagnosis and then seen every 6–12 months, or more often if needed. The list below includes many of the common problems, but others may arise. If you have any concerns about your child's health, please speak with your family doctor (GP) or paediatrician.

Possible health problems	Management recommendations
Seizures (epilepsy)	• Anti-seizure medications (ASM) can improve seizure control for some children • The response to ASMs appears to vary between different SCN2A types. This means that treatments that appear to benefit children with certain clinical features (phenotypes) may not help children with other types of epilepsy. Treatment choice is critical because some medications can make seizures worse • Seek the opinion of a paediatric neurologist through the usual referral pathways • Further information about specific treatments is available at Human Disease Genes
Developmental delay/ intellectual disability	• Early intervention including speech therapy, occupational therapy and physiotherapy • Special education programs
Reflux and/or constipation	• Review and management by paediatrician
Movement disorder: uncontrolled muscle tightening (dystonia), periods of poor coordination or balance (episodic ataxia)	• Seek the opinion of a paediatric neurologist through the usual referral pathways
Autism spectrum disorder/ challenging behaviours	• Diagnosis and management by paediatrician if needed
Difficulties going to and staying asleep	• Review by paediatrician; consider referral to sleep physician if severe symptoms

Detailed management recommendations for healthcare professionals can be found at **Human Disease Genes**.

Resources, support and connecting with others

You may find it helpful to connect with other people who have personal experience of day-to-day life with a child who has an *SCN2A*-related condition or a similar genetic condition. You can make these connections through:

- Social media (e.g. closed Facebook groups such as **SCN2A – Australia, NZ and Asia**)
- Condition-specific groups (e.g. **SCN2A Australia**)
- Groups for individuals with common symptoms that may have many different causes (e.g. intellectual disability, hearing loss, autism).
- Umbrella groups (e.g. **Genetic Alliance Australia**)

SCN2A Australia can provide support for families, as well as information about specialised clinics or expert clinicians for *SCN2A*-related conditions. Many organisations (e.g. **Carers NSW** and **Kindred**) can also offer general advice and support in caring for a family member with long-term needs.

It is important to know that you are not alone on this journey

More information about *SCN2A*-related conditions

You can find further information about *SCN2A*-related conditions by following the links below.

- UNIQUE: **SCN2A related conditions**
- **SCN2A Australia**
- Human Disease Genes: **SCN2A**

For more information about genetic conditions and to find your local Clinical Genetics services, visit the **NSW Centre for Genetics Education**. More information about childhood epilepsy is available at the **Paediatric Epilepsy Network NSW** website.

Family planning

Genetic conditions are sometimes passed from a parent to their child. Even if the same variant was not found in one of the parents, it is still possible to have another child with the same condition. The chance of this happening is about 1%.

If you are thinking about having more children, it is recommended that you talk with your local **Clinical Genetics service**. Some people may choose to have **genetic testing** before or during a pregnancy. Specialised health professionals such as **genetic counsellors** can advise you on your options.

You can also speak with your GP about options for **reproductive genetic carrier screening**. When planning a family, it is best to explore your options before becoming pregnant.

Research, registries and clinical trials

Some people with genetic conditions are able to participate in research, which may be of benefit to your child. This may investigate how a particular gene change causes health problems or it may be a **clinical trial** testing new treatments.

Another way to contribute to research is to join the **SCN2A natural history study**. A natural history study collects information about many aspects of a health condition. This includes the course of the condition over time, as well as information about the people with the condition and their families, including where they live and their family history.

Sharing information about your child's signs and symptoms through registries such as **FaceMatch** and **IAMRARE** can help build further knowledge about this condition.

Information about current clinical trials can be found by searching the **Australian Clinical Trials** website or the international databases **ClinicalTrials.gov** or **EudraCT**. Some researchers in Australia are currently investigating whether specific medicines can help with *SCN2A*-related conditions. You can contact **SCN2A Australia** or the Neurology Department at Royal Children's Hospital Melbourne if you would like more information on these clinical trials.

To print more copies of this fact sheet and access links to the underlined topics, go to **https://www.genetics.edu.au/SitePages/SCN2A.aspx**

The recommendations in this fact sheet were current at the time it was written. This fact sheet should not replace a consultation with a specialist healthcare professional.

Version 1. June, 2022.