

Therapy/Treatment options for SCN2A

In short...

- No treatment is currently available that can cure an individual with an SCN2A-related condition.
- Individuals with SCN2A usually need to see many different healthcare professionals at different times.
- Management by a **neurologist** is recommended for individuals with epilepsy and/or movement disorders.
- **Early intervention programs, occupational therapy** and **speech therapy** are valuable for individuals with developmental delay / intellectual disability and/or autism spectrum disorder (ASD).
- The choice of anti-seizure medication is important. Certain medications can make seizures worse in some individuals

Individuals with SCN2A-related conditions usually receive many different types of treatment, including antiseizure medications, early intervention programs, and occupational or speech therapy.

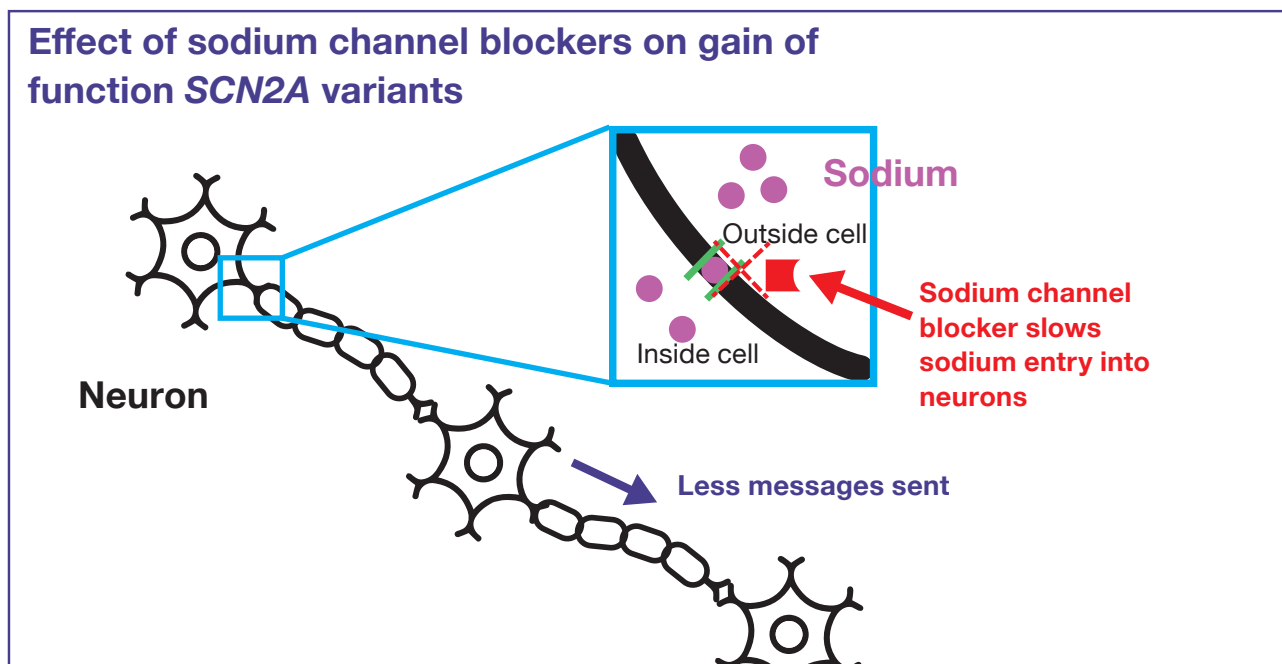
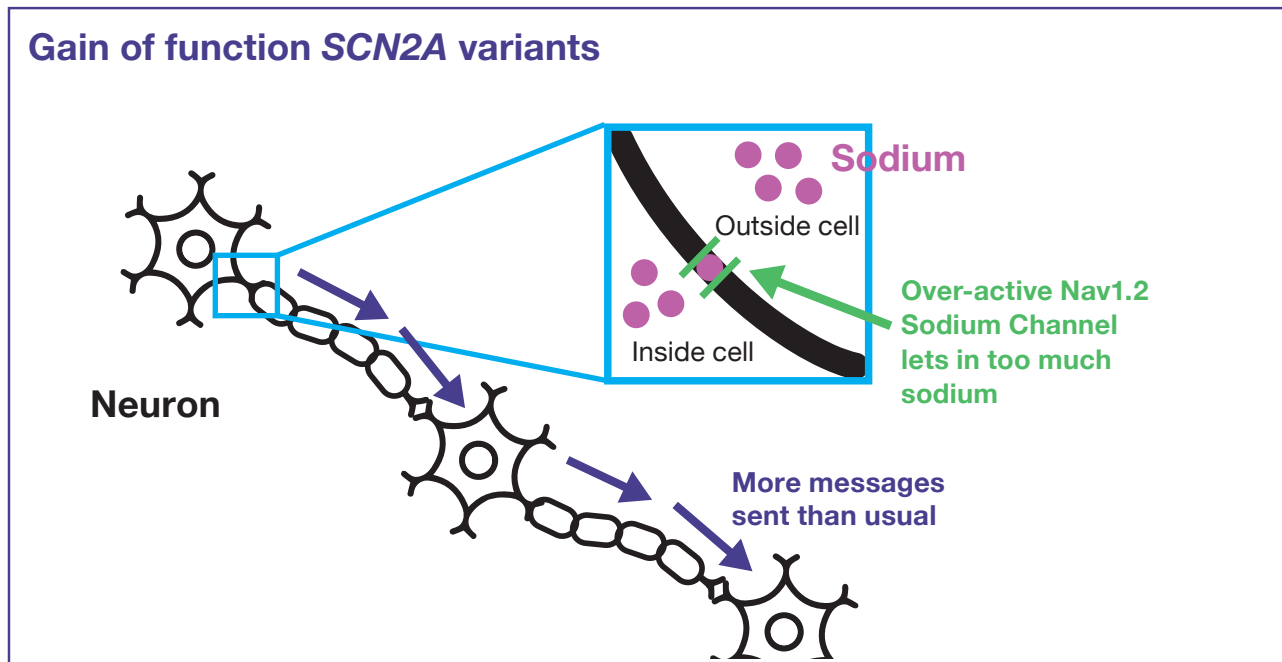
Treatment of epilepsy

Anti-seizure medications ^{1,2}

- The type of anti-seizure medications used will depend on the type and severity of the seizures.
- As no single anti-seizure medication has been shown to be effective in all individuals with SCN2A, many different medications may need to be tried. A combination of anti-seizure medications may be needed.
- Response to anti-seizure medications seems to vary between different SCN2A types. Medicines that are effective in some individuals may not work in others.

Sodium channel blockers ³⁻⁷

- The anti-seizure medicines known as **sodium channel blockers** improve seizure control in many individuals with epilepsy that starts before the age of 3 months.
- These medications are particularly beneficial in individuals with so-called '**gain of function (GOF)**' *SCN2A* variants (see Information Sheet #2 "What does gain of function and loss of function mean?")
- In these individuals, the sodium channel blocker works by stopping the overactive Nav1.2 sodium channel. This slows down the messages that are sent between brain cells (neurons).



- Sodium channel blockers do not work in all individuals with epilepsy. When there is a 'loss of function (LOF)' *SCN2A* variant, blocking the already-underactive sodium channel can make seizures worse.

Other options¹

These may be considered when medication is not working.

Possible treatments include:

- Implantable devices e.g. vagus nerve stimulators
- Diet therapy eg the **ketogenic diet**.

Management of other symptoms¹⁻⁶

- No *SCN2A*-specific treatments for developmental delay / intellectual disability or behavioural symptoms are currently known.
- Individuals with developmental delay / intellectual disability or ASD are supported with standard interventions and treatments, including:
 - o Early developmental and behavioural interventions, such as specialist schooling, community programs, and social skills support
 - o Speech therapy, occupational therapy and physiotherapy.
- Individuals are monitored for other problems such as constipation or reflux, sleep disorders and hypotonia. If identified, these are treated using standard approaches.

Gene-based treatments in development⁸⁻¹⁰

- Another type of treatment being investigated for *SCN2A* is **gene therapy**.
- Gene therapy for *SCN2A* specifically targets the *SCN2A* gene. It may target specific changes (variants) in the *SCN2A* gene, or specific types of variants. This means that gene therapy that works for one person may not work for another.
- Gene therapies may:
 - o Turn the *SCN2A* gene 'up', so that more Nav1.2 channels are made.
 - o Turn the gene 'down', so fewer Nav1.2 channels are made.
 - o Tell the *SCN2A* gene to make a full-size Nav1.2 channel, instead of a shortened version.
- For more information about gene therapy, see Information Sheet #6 "Gene Therapy".

More information

- Children's Hospital of Philadelphia
- Centre for Genetics Education
- Human Disease Genes



References

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