We are focused on supporting **families** and finding a **cure**

Have you just received a diagnosis of mutation of the gene SCN2A?

You are not alone!

DONATE NOW!

DONATIONS:
SCN2A ITALIA Famiglie in Rete APS
IBAN: IT10G0623012141000015281975
PayPal.Me/SCN2aItalia

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SCN2AITALIA
FAMIGLIE IN RETE

Providing support to Families with children/adults with

SCN2A related conditions

Who we are

SCN2A ITALIA Famiglie in Rete is an organisation created in 2021 by parents and siblings of children/adults diagnosed with rare forms of Epilepsy and Autism as a result of a change in the SCN2A gene.

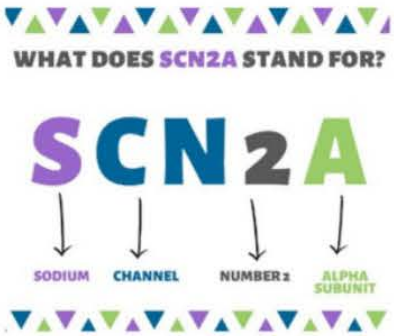
"Families" (in italian: "Famiglie") is part of our name for a reason. Rare and devastating, SCN2A-related disorders affect the entire family. We strive every day and in every way to improve the lives of not only the patients, but the entire family.
In May 2022 we were included in the National Registry for Organisations operating in the Voluntary Sector (RUNTS).

Our **MISSION** is to accelerate research, build community and advocate to improve the lives of those affected by SCN2A-related disorders.

Our **VISION** is a world with effective treatments and cures for all SCN2A-related disorders.

The gene SCN2A

The SCN2A gene is located on the long (q) arm of chromosome 2 at position 24.3. SCN2A encodes instructions to make a protein in the brain called a sodium channel which plays a key role in a cell's ability to generate and transmit electrical signals. Pathogenic variants that affect the SCN2A sodium channel impair the flow of sodium ions in the brain.



Most children affected by an SCN2A mutation have a wide range of disorders:

- Benign familial neonatal /infantile seizures
- Epilepsy
- Epileptic encephalophaties
- Developmental delay
- Hypotonia (floppiness of the body)
- Autism
- Gastrointestinal problems
- Sleep Problems

Our targets

Support

We provide daily support to parents and siblings of children/adults with SCN2A related conditions

Research

We promote research projects through the help of our Scientific Commitee to strengthen scientific knowledge and enhance the professional development of researchers.
Our goal is to find a CURE.

Networking

We closely collaborate with other associations, foundations both nationally and internationally. . We wish to create a strong network to achieve our common goals.

Awareness and Resources

We strongly believe in increasing awareness and advocate for the need and utility of genetic testing.

Although SCN2A is one of the leading single-gene causes of neurodevelopment disorders, it is significately under-diagnosed.

We strongly wish to **improve quality of life** of those affected by this ultra-rare condition.