FAQ for The Cute Syndrome Foundation Global SCN8A Survey Series

1. What is a Patient Registry?

A patient registry is a collection of standardized information about a group of patients who share a condition and is used for a variety of purposes such as conducting natural history studies and supporting disease specific clinical trial recruitment.

2. What is a Natural History Study?

A natural history study is a study designed to track the course of a disease over time and includes people who have a specific medical condition or disease and those who are at risk of developing such. This method of research explores the disease in a comprehensive way and identifies demographic, genetic, environmental, and other variables that correlate with the disease and its outcomes. Natural history studies have many potential uses such as patient care best practice developments and clinical trial recruitment.

3. What is a Research Study Sponsor?

An individual, company, institution, or organization that takes responsibility for choosing appropriately trained and experienced researchers as well as the initiation, management, and/or financing of a clinical trial. The study sponsor ensures that the study is conducted in a reputable manner and upholds regulations as they apply to the study.

4. What is a Principal Investigator?

The Principal Investigator is the research group leader or, the person with the primary responsibility for the design and conduct of the research project or study.

5. What is an Institutional Review Board (IRB)?

Any board, committee, or other group formally designated by an institution or investigator to review, approve the initiation of, and to conduct periodic review of research involving human subjects. The primary purpose of such review is to assure the protection of the rights and welfare of the human subjects. Also known as Ethics Committee (EC).

6. What is the purpose of The Cute Syndrome Foundation Global SCN8A Survey Series**?**

One of the most important purposes of The Cute Syndrome Foundation Global SCN8A Survey Series is to bring the SCN8A community together and collect data, which could be used to create therapeutics and improve the quality of life for patients. Some other goals of The Cute Syndrome Foundation Global SCN8A Survey Series are to:

- Conduct a prospectively planned natural history study that will result in the most comprehensive understanding of SCN8A and its progression over time.

- Characterize and describe the SCN8A population as a whole.
- Assist the SCN8A community with the development of recommendations for standards of care.
- Assist researchers studying the pathophysiology of SCN8A
- Assist researchers studying interventional outcomes.
- Support the design of clinical trials for new treatments.

7. What types of data will be collected in The Cute Syndrome Foundation Global SCN8A Survey Series**?**

The data collected is uniform and includes but is not limited to

- Socio-demographics
- Medical and diagnostics
- Treatment and disease progression
- Management of care
- Quality of life

8. How is the data collected?

Data is collected through a secure web-based system developed by the National Organization for Rare Disorders (NORD), an independent non-profit committed to the identification, treatment, and cure of all 7,000 rare diseases. Study participants respond to questions grouped within a series of surveys developed per study standards and in collaboration with disease specific experts.

9. Who is a study participant?

A study participant is the individual who takes part in a research study and whose information is collected for that research. Study participants may consent to enter and share their own personal data.

10. Who is a reporter/respondent?

A reporter/respondent is an individual who completes the surveys on behalf of the patient/study participant, when they are unable to do so on their own behalf.

11. What is a legally authorized representative (LAR)?

An individual who is authorized under applicable law to consent, on behalf of a prospective subject, to the subject's participation in the clinical trial. The LAR may be a parent, grandparent, caregiver, or guardian who has the legal authority to grant consent on behalf of another who is eligible to participate in research. When a LAR acts on behalf of a study participant, he/she is considered to be the reporter/respondent in the research.

12. What is an Informed Consent?

The Office for Human Research Protections (OHRP) states that, "… the informed consent process is the critical communication link between the prospective human subject and an investigator beginning with the initial approach of an investigator to the potential subject (e.g. through a flyer, brochure, or any advertisement regarding the research study) and continuing until the completion of the research study. […] The informed consent process involves three key features: (1) disclosing to potential research subjects' information needed to make an informed decision; (2) facilitating the understanding of what has been disclosed; and (3) promoting the voluntariness of the decision about whether or not to participate in the research."[1]

13. Who can join the study?

This study is open to anyone who has a **SCN8A diagnosis.**

14. Is there a cost to participate?

There is no cost to the patient to join this study. The Cute Syndrome Foundation absorbs the cost of the registry for its members.

15. How long will this study last?

This registry will be open for at least five years with the option to renew registration. There is no date of termination or closure at this time.

16. Can data be collected worldwide?

The patient registry uses an online platform which allows participants to contribute data from anywhere in the world. Individuals from other countries who enter data into the registry should be aware that data and privacy laws are different in the U.S. from other countries. This U.S. based registry will protect data and privacy according to U.S. requirements.

17. Who is NORD – the National Organization for Rare Disorders?

The National Organization for Rare Disorders (NORD®), and independent nonprofit, is leading the fight to improve the lives of rare disease patients and families. We do this by supporting the rare community, its people and organizations. We work together to accelerate research, raise awareness, provide valuable information and support, and drive public policy that benefits the estimated 25-30 million Americans impacted by rare diseases. Learn more about NORD at https://rarediseases.org/.

18. Where is the data stored?

The data is stored on NORD's registry platform system which adheres to industry standards regarding security protections.

19. Is the data safe?

Yes, the data is safe. The registry follows strict government guidelines to assure patient information is protected. The platform is served over HTTPS, which provides traffic encryptions so as to prevent eavesdropping and man-in-the-middle attacks. Communications between the registry platform application server and the database are also encrypted.

20. How is the Patient Registry maintained?

The registry is maintained by NORD who hosts the registry on its cloud-based Platform and provides oversight and ongoing support of the system. **The Cute Syndrome Foundation** provides the day-to-day management of their patient registry, including the development and adherence to the study procedures.

21. Who is The Cute Syndrome Foundation?

The Cute Syndrome Foundation raises awareness of SCN8A mutations, funds the dedicated and talented scientists researching SCN8A, and supports the families around the world who are affected by this disorder.
Learn more about The Cute Syndrome at https://www.thecutesyndrom.com

22. Who owns the data?

The identifiable and de-identifiable data are owned by the study sponsor, The Cute Syndrome Foundation. **The Cute Syndrome Foundation** decides how and with whom to share the data. A subset of the de-identified data collected across the NORD Registry Platform is available to NORD to support cross disease analysis and advocacy activities to members of the rare disease community as a whole.