FAQ for SPS Global Registry

1. What is a Patient Registry?

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

2. What are the Objectives of the SPS Global Registry?

The primary objective of the SPS Global Registry is to conduct a natural history study (NHS) aimed at understanding the progression and overall course of Stiff Person Syndrome (SPS). By gathering detailed data, the Registry seeks to characterize patients with SPS, documenting their symptoms and identifying common patterns. This initiative also aims to provide critical insights into the patient burden of SPS, including its impact on quality of life and the long, often difficult diagnostic journey faced by those with the condition. Additionally, the Registry will collect valuable information on therapeutic interventions and their outcomes, helping to shape future treatment approaches.

Beyond these goals, the SPS Global Registry will serve as a critical resource by establishing a pool of patients diagnosed with SPS who may be eligible to

participate in ongoing and future research. An integral component of this effort is the creation of a biorepository linked to the Registry, which will advance research by providing biological samples for study. Moreover, by encouraging international collaboration, the registry will foster partnerships and promote global efforts to advance SPS research, ultimately striving to improve patient care and outcomes.

3. What is a Natural History Study?

A natural history study is a study designed to track the course of a disease over time. It includes people who have a specific medical condition or disease. It may also include those who are at risk of developing the condition/disease. This type of research identifies demographic, genetic, environmental and other information that may be common within the disease and its outcomes. A natural history study can also show the differences in symptoms and changes over time that are seen in different people with the same disease. Natural history studies often aim to find unknown similarities within the disease population. They have many potential uses such as patient care best practice development and clinical trial recruitment. Data for natural history studies are often collected via patient registries.

4. Who oversees the SPS Global Registry?

The Stiff Person Syndrome Research Foundation (SPSRF) is the Registry's research study sponsor and is responsible for overseeing the SPS Global Registry.

5. What is a Research Study Sponsor?

A Research Study Sponsor is an individual, company, institution, or organization. They are responsible for:

- Choosing appropriately trained and experienced researchers to conduct the research study;
- Initiating and managing the research study;

- Ensuring funding for the research study;
- Ensuring that the research study is conducted in a reputable, ethical manner and upholding regulations as they apply to the study; and
- Maintaining regulatory compliance and selecting an Institutional Review Board (IRB) that will oversee the conduct of the study.

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

An IRB is a board formally designated by an institution or investigator to review, approve the initiation of, and conduct periodic review of research involving people. The primary purpose of such an assessment is to ensure the protection of the rights and welfare of the participants in the study. This is also known as an Ethics Committee (EC) or Research Ethics Board (REB in Canada).

7. Who can join the study?

This study is open to anyone who has a confirmed diagnosis of SPS by a physician. A confirmed diagnosis consists of clear documentation in a patient's medical records, such as a progress note, stating the patient has an SPS diagnosis. This documentation will need to be uploaded into the Registry.

8. Is there a cost to participate?

There is no cost to participate in the Registry.

9. What types of data will be collected in the SPS Global Registry?

The data collected includes details about:

- Socio-demographics
- Medical History
- Women's Health
- Lifestyle
- Family Medical History
- SPS Diagnosis and Testing

- Medical Uploads
- SPS Symptoms
- SPS Treatments
- Quality of life

10. How is the data collected?

Data is collected through a secure web-based application (that can be accessed by computer, tablet or phone) developed by the National Organization for Rare Disorders, Inc. (NORD®), Study participants respond to questions grouped within a series of surveys developed per study standards and in collaboration with disease specific experts.

11. Who is NORD – the National Organization for Rare Disorders, Inc.?

NORD, an 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 drive public policy that benefits the estimated 25-30 million Americans impacted by rare diseases. Learn more about NORD at https://rarediseases.org/.

12. Is the data secure?

The Registry follows strict government guidelines to ensure patient information is protected. The platform is served over HTTPS, meaning the data is encrypted when sent from the user's browser to the NORD servers. The data is also kept encrypted in the NORD database. Communications between the registry platform application server and the database are also encrypted. As with any information you provide electronically, there is a rare chance that your privacy could be compromised. However, the Registry and the security measures minimize the possibility of this occurring.

13. Where is the data stored?

NORD stores Sponsor and Participant Registry Data on NORD encrypted servers and/or encrypted servers of third-party vendors hosted in Canada. Regular back-up at commercially acceptable intervals is provided. These servers meet industry standards and are compliant with US and international regulations, including GDPR.

14. What are the GDPR considerations?

For individuals living outside the United States who choose to share information about themselves, the same protections for privacy and confidentiality are offered as in the United States. Residents of the European Union and Switzerland have additional particular rights related to personal information. This information is disclosed within the informed consent document. If an individual signs this document, they acknowledge that they are disclosing information that would otherwise be private. Privacy laws in an individual's country may have different protections than those provided in the United States.

Registry participants who are residents of the European Union and Switzerland are entitled to:

- Request to obtain access to and rectification or erasure of personal data;
- Receive personal data in a portable, readily-accessible format;
- Restrict or withdraw permission for the processing of personal information; and
- Lodge a complaint with an appropriate supervisory authority.

15. Can data be collected worldwide?

The 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.

16. Who owns the data?

The study sponsor owns the study data. The SPSRF decides how and with whom to share the data. NORD staff will have access to the data for activities related to support and maintenance of the Platform and will collect Platform-wide participation statistics. The specifics will be outlined in your informed consent.

17. Who will have access to Protected Health Information (PHI)?

All data, including those with PHI, will be stored in a password protected secure server. Access to PHI will be limited to:

- Approved members of the SPS Global Registry research team;
- NORD staff, in cases where technical support is needed and with the permission of registry staff; and
- With agreement from the Sponsor, NORD may conduct IRB-approved, cross-disease research using registry data.

In all cases, your privacy will be protected.

18. Health Information How will the data be shared with researchers?

The Sponsor and the PI in consultation with the co-investigator and/or members of the Registry Steering Committee will evaluate all requests for data from researchers. Researchers will only be provided with the minimum data necessary to accomplish their research study goals. Data containing PHI will only be shared if the research cannot be done without it. The researchers will be required to sign a Confidentiality Agreement in which they promise not to share data with anyone else.

19. What is a Registry Steering Committee?

A Registry Steering Committee is a group of experts that includes patients, caregivers, clinicians, researchers, and data specialists who advise the sponsor on developing, implementing, and managing the Registry.

20. What is a Principal Investigator?

The Principal Investigator (PI) is the person with the primary responsibility for the design and conduct of the research project or study. The PI is responsible for oversight of all aspects pertaining to the conduct of the Registry, its staff and the research on the data contained within.

21. Who is a Study Participant?

A Study Participant is the individual about whom information is entered into the Registry. In the case of an independent person of legal age, this individual will consent for and enter information about themself. If an individual is not of legal age or is an adult who requires someone to act on their behalf, a person (Caregiver/LAR, see below) who is legally responsible for their health care will provide consent and enter information about the Study Participant.

22. Who is a Study Partner?

A Study Partner is a close relative or trusted friend who can assist the Study Participant, who is able to consent and answer questions for themself, in completing the surveys when they need help due to SPS related symptoms or other medical conditions. Their role as a Study Partner is to read the surveys to the Study Participant and enter their responses into the surveys exactly as directed by them. Using this assistance may add additional risk of exposure of your personal information to the person physically entering data for you, and a risk that the data you relay may be entered incorrectly.

23. What is a Legally Authorized Representative (LAR)?

An LAR is someone who is authorized under applicable law to consent and enter data in the registry on behalf of another individual. The LAR may be a

parent, grandparent, spouse, caregiver, or guardian as long as they have the legal authority to grant consent on behalf of that individual. An LAR will sign up on the IAMRARE platform with a Caregiver account. When an LAR acts on behalf of a study participant, they are considered to be the reporter in the research.

24. What is a Designated Representative?

A Designated Representative is a legal adult who was the caretaker of an individual with SPS who passed away. This may be a spouse, parent, sibling, offspring, close relative, close friend, guardian and/or significant other of this individual. This person must have had knowledge of and participated in the deceased's medical care. These individuals are permitted to enter retrospective data on their behalf.

25. What is an Informed Consent Form (ICF)?

An ICF is a document that provides potential participants with key information about the Registry. This document helps potential participants to make an informed decision whether to join or not. Information will include topics such as: the risks and benefits of the research project, use of data, and participant privacy. If they choose to join the study, participants are required to electronically sign the ICF. This indicates that they agree to the terms as described before entering data into the Registry or responding to surveys.

26. After consenting, can I withdraw from the study?

Participants can withdraw from the study at any time. However, information already used for research before the participant changed their mind cannot be retrieved.