FAQ for the Gorlin Syndrome Alliance Patient 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 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. Who is the Gorlin Syndrome Alliance?

The Gorlin Syndrome Alliance is a national 501 (c) 3 organization who thoughtfully supports, comprehensively educates, and aggressively seeks treatments and a cure for Gorlin syndrome, its manifestations and sporadic BCCs. The organization has a strong desire to build upon and strengthen our alliance between patients, medical professionals, the pharmacological industry, professional staff, and lay leadership while continuing our commitment to provide strong emotional and informational support to our members.

5. Who is NORD® – the National Organization for Rare Disorders, INC.?

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 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/.

6. 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.

7. 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).

8. What is the purpose of the Gorlin Syndrome Alliance Patient Registry?

One of the most important purposes of the Gorlin Syndrome Alliance Patient Registry is to bring the Gorlin syndrome 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 Gorlin Syndrome Alliance Patient Registry are to:

- Conduct a prospectively-planned natural history study that will result in the most comprehensive understanding of Gorlin syndrome and its progression over time.
- Characterize and describe the Gorlin syndrome population as a whole.
- Assist the Gorlin syndrome community with the development of recommendations for standards of care.
- Assist researchers studying the pathophysiology of Gorlin syndrome.
- Assist researchers studying interventional outcomes.
- Support the design of clinical trials for new treatments.

9. What types of data will be collected in the Gorlin Syndrome Alliance Patient Registry?

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

10. How is the data collected?

Data is collected through a secure web-based system developed by the National Organization for Rare Disorders, Inc. (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.

11. 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.

12. 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.

13. 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, they are considered to be the reporter/respondent in the research.

14. 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]

15. Who can join the study?

This study is open to anyone who has a Gorlin syndrome diagnosis.

16. Is there a cost to participate?

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

17. 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.

18. 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.

19. Where is the data stored?

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

20. 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.

[1] Informed Consent FAQs: https://www.hhs.gov/ohrp/regulations-and-policy/guidance/faq/informed-consent/index.html. Accessed Feb. 9, 2021.

21. Who owns the data?

The identifiable and de-identifiable data are owned by the study sponsor, Gorlin Syndrome Alliance. Gorlin Syndrome Alliance] decides how and with whom to share the data. A subset of the pseudonymized 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. Participants are able to withdraw from the study at any time, however, the researchers may still use the information that they have collected prior to changing your mind in order to complete the research that has already started. Information that has already been shared with the RDCA-DAP or sent to a researcher for a specific study prior to your request for removal cannot be retrieved or removed.

22. What is a registry Advisory Board?

A Registry Advisory Board committee, that may include scientists, doctors, and patient advocates, will be assembled to oversee the conduct of the study. The Advisory Board will review aggregate registry data and the use of this registry, ensure proper evaluation of protocols requesting to use registry data and/or contact registry participants, and will review any protocol or confidentiality deviations on a case by case basis and ensure that any such deviations are reported to the IRB.

23. 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. Gorlin Syndrome Alliance provides the day-to-day management of their patient registry, including the development and adherence to the study procedures.