



FAQ for IAMRARE Natural History Study 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 (NHS)?

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?

The National Health Service defines a study sponsor as, “the individual, company, institution or organization, which takes on ultimate responsibility for the initiation, management [...] of and/or financing [...] for that research.”¹ 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)?

According to the Mayo Clinic an IRB is, “a specifically constituted review body established to protect the rights and welfare of human subjects recruited to participate in biomedical or behavioral/social science research.”² An institutional review board is a group of people who are responsible for protecting the rights and welfare of people who participate in studies.

¹ Roles and Responsibilities: Sponsor. National Health Service Health Research Authority Web site. <http://www.hra.nhs.uk/resources/before-you-apply/roles-and-responsibilities/sponsor/> Accessed November 7, 2016.

² Institutional Review Board (IRB) Definition of Terms. Mayo Clinic Web site.

<http://www.mayo.edu/research/institutional-review-board/definition-terms> Accessed November 7, 2016.

6. What is the purpose of the IAMRARE NHS Patient Registry Program?

One of the most important purposes of the IAMRARE NHS registry program is to bring rare disease communities together and collect data which could be used to create therapeutics and improve the quality of life for patients. Some other goals of the registry program are to

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

7. What types of data can be collected in a Patient Registry?

The data collected 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)?

A legally authorized representative is an individual who, under law, has the ability to act on behalf of another person (such as a minor study participant). The LAR may be a parent, grandparent, caregiver who has the legal authority to grant consent on behalf of another who has been invited 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 study.

12. Can data be collected worldwide?

The patient registry uses an online platform which allows participants to contribute data from anywhere in the world.

13. Where is the data stored?

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

14. 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.

15. Who owns the data?

The identifiable and de-identifiable data are owned by the study sponsor. The study sponsor 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.

16. 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 study sponsor provides the day-to-day management of the patient registry, including the development and adherence to the study procedures.