FAQ for KAT6A Patient Registry Natural History Study

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 longitudinal study?

According to the British Medical Journal, a longitudinal study follows subjects, "…over time with continuous or repeated monitoring of risk factors or health outcomes, or both. […] Most longitudinal studies examine associations between exposure to known or suspected causes of disease and subsequent morbidity or mortality."[1] Longitudinal research projects can extend over years or even decades.

4. 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.

5. 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.

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

[1]Informed Consent FAQs: https://www.hhs.gov/ohrp/regulations-and-policy/guidance/faq/informed-consent/index.html. Accessed May 15, 2017.

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).

7. What is the purpose of the KAT6A Patient Registry?

One of the most important purposes of the KAT6A is to bring the KAT6A 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 the KAT6A Patient Registry are to:

- Conduct a prospectively-planned natural history study that will result in the most comprehensive understanding of KAT6A Syndrome and its progression over time.
- Characterize and describe the KAT6A Syndrome population as a whole.
- Assist the KAT6A Syndrome community with the development of recommendations for standards of care.
- Assist researchers studying the pathophysiology of KAT6A Syndrome.
- Assist researchers studying interventional outcomes.
- Support the design of clinical trials for new treatments.

8. What types of data will be collected in the KAT6A 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

9. 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.

10. 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.

11. 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.

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

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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.

13. 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]

14.Who can join the study?

This study is open to anyone who has a KAT6A Syndrome diagnosis.

15. Is there a cost to participate?

There is no cost to the patient to join this study. The KAT6A Foundation absorbs the cost of the registry for its members.

16. How long will this study last?

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

17. 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. For persons living outside the U.S. who choose to share information about themselves, the same protections for privacy and confidentiality are offered as in the U.S.

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

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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.What are some risks of the study?

There are no risks of physical harm associated with participation in the study. Participation in the KAT6A Patient Registry does involve the potential risks of a breach of confidentiality of medical information and associated privacy of participants. Such risks will be minimized by ensuring adherence to applicable regulations and data security measures and by performing the following: (1) removing direct participant identifiers from information and data shared or released from the registry; (2) limiting access to linking codes assigned to the KAT6A Patient Registry information; 3) and limiting access to information contained within the KAT6A Patient Registry to registry investigators and researchers approved by the Advisory Board, and (4) Maintaining the Privacy and Confidentiality of Registry information.

21. Who owns the data?

The identifiable and de-identifiable data are owned by the study sponsor, The KAT6A Foundation. The KAT6A 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.

22.Who will be able to use this data?

One of the goals of this registry is to gather and disperse KAT6A Syndrome information as quickly and securely as possible. In agreement with the standards set by NORD and the IRB, we plan to share *de-identified* data with *any* researcher who asks for it for a legitimate reason. The KAT6A Foundation in collaboration with the KAT6A Foundation/Registry Advisory Board (comprised of Clinical and research experts, and family advocates will review requests for access to de-identified data from researchers. Investigators wanting to use the registry or contact participants will need to apply to the KAT6A Foundation Advisory Board for access. The application will require information concerning: Principal Investigator, aims and hypotheses of the proposed research, and where the research will be performed, and how the research will be funded. The KAT6A Foundation Advisory Board will review and approve applications based on study quality, potential, and value to KAT6A Syndrome.

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. **[Organization name]** provides the day-to-day management of their patient registry, including the development and adherence to the study procedures.

24.Who is The KAT6A Foundation?

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The KAT6A Foundation was created in 2017 by parents of children identified with a mutation in the KAT6A gene. We are the first ever 501(c)(3) nonprofit organization founded to support the international KAT6A syndrome community. Learn more about The KAT6A Foundation at https://www.kat6a.org/

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

NORD, a 501(c)(3) organization, is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. NORD was founded by patients and families who marshaled grass-roots efforts to secure the passage of the Orphan Drug Act in 1983. Today, NORD represents the united voice of more than 250 rare disease-specific groups and thousands of patient advocates. Together, we are committed to the identification, treatment and cure of rare disorders through programs of advocacy, education, research and patient support services. Learn more about NORD at https://rarediseases.org/.

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