FAQ for Albinism International Databank (AID)

1. What is the purpose of the Albinism International Databank (AID)?

The purpose of the Albinism International Databank is to bring the Albinism/HPS/CHS community together and collect data.

Some of the goals of the AID are:

1. Establish a carefully defined cohort (group) of people with Albinism.
2. Analyze and describe the characteristics of the condition and the number of people affected worldwide by looking at the information in the AID with help from experts in the field.
3. Help our community develop recommendations on how to treat the condition based on the information collected and advice from our experts
4. Find out how many and how severe different Albinism symptoms are in the non-HPS/CHS community.
5. Learn if there are any HPS/CHS symptoms in the Albinism community by looking at the information in the AID.
6. To identify which tests are being used the most in diagnosing your condition around the world.
7. Find out how many and how severe different HPS/CHS symptoms are in the HPS/CHS community by looking at the information in the AID.
8. To identify people with Albinism/HPS/CHS who might be willing to take part in other research studies or clinical trials. You will be able to choose whether you want to hear about these other studies.
9. Provide a secure and user-friendly platform for you to share your medical history and experiences to be stored for future research.
10. The AID will be a resource for researchers studying these conditions by giving them access to your de-identified data (including biospecimens when available), to aid in the development of new treatments.
11. Understand the impact of having Albinism/HPS/CHS has on mental health and activities of daily living by examining information in the AID.
12. Find out how often and whether other medical conditions occur by looking at the information in the AID.
13. Upload health information from your wearables to collect patient recorded outcomes when this becomes available in the platform.

2. 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.

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. 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®), (learn more about NORD in question 24). Study participants respond to questions grouped within a series of surveys developed per study standards and in collaboration with disease specific experts.

5. What types of data will be collected in the AID?

The data collected includes but is not limited to:

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

6. 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 study. They are also responsible for the initiation and management of a research study. Additionally, the sponsor is responsible for the costs associated with conducting a registry study. They ensure that the study is conducted in a reputable, ethical manner and upholds regulations as they apply to the study. The sponsor of this registry is the Hermansky-Pudlak Syndrome Network (HPS Network) Inc. and the Collaborating Organization is the National Organization of Albinism and Hypopigmentation (NOAH).

7. Who are the HPS Network and NOAH?

The HPS Network is a 501c3 non-profit that provides education and vital support programs to individuals and families with Hermansky-Pudlak syndrome while striving for improved care and innovative research on our journey to cure.

To achieve this goal, the HPS Network gathers and disseminates information, promotes

awareness, funds research, and provides support to our members. This keeps us actively developing educational materials, pamphlets, presentations, and articles to help both families and professionals understand the needs created by this syndrome. We maintain a contact and symptom registry to assist with informing and networking individuals, recruiting for research trials, and facilitating frequent communications. We have a toll-free number to provide access to support with a bilingual office ready to help.

NOAH serves the albinism community by providing information and support. We envision a world where people with albinism are empowered to be fully-functioning members of society, where barriers and the stigma of difference no longer exist, and where people with albinism have a quality of life that is rewarding, dignified and fulfilling. NOAH's mission is to act as a conduit for accurate and authoritative information about all aspects of living with albinism and to provide a place where people with albinism and their families in the U.S. and Canada can find acceptance, support, and fellowship.

8. 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.

9. 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.

10. 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.

11. What is a Designated Representative?

A Designated Representative is a legal adult who was the caretaker of an individual who passed away from Albinism/HPS/CHS. 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 medical care of the deceased. These individuals are permitted to enter retrospective data on their behalf.

12. 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.

13. After consenting, can a Participant choose to stop participating in the study?

Participants are able to withdraw from the study at any time. However, researchers may still use the information that they have collected prior to the participant changing their mind. Information that has already been shared with researchers prior to withdrawal cannot be retrieved or removed.

14. 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 assure 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).

15. What is a Registry Advisory Board?

A Registry Advisory Board is a committee that may include scientists, doctors, and patient advocates. They oversee the conduct of the study. The board advises on the development of surveys and reviews combined registry data and the use of this registry. They will ensure proper evaluation of all research requests for use of the registry data. They will also review any protocol or confidentiality deviations and ensure that any such deviations are reported to the IRB.

16. Who can join the study?

This study is open to anyone who has an Albinism/HPS/CHS diagnosis and meets the study inclusion criteria for participation.

17. Is there a cost to participate?

There is no cost to the patient to join this study.

18. Is there a payment for participating?

It should not cost you anything to participate in the AID, other than the costs of internet access. You will not be paid for the information you provide.

Periodically, and at the discretion of the Sponsor, you may elect to receive gifts of nominal value (for example, pens, posters, promotional apparel/jewelry, donation in your name to a non-profit organization or gift cards of $25.00 or less, etc.)

Your information will only be used for research, but it is possible that the research could lead to the development of a commercial medical product. Should this happen, you should not expect to be paid.

19. How long will this study last?

A registry on the IAMRARE platform will typically be open for at least five years. Participants will be asked to return to the registry periodically to update their information.

20. 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.

21. 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.

22. 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.

23. Is the data safe?

The registry follows strict government guidelines to assure patient information is protected. The platform is served over HTTPS, which means that the data is encrypted when being 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 very rare chance that your privacy could be compromised. However, the registry and the security measures minimize the chance of this occurring.

24. Who owns the data?

The study data are owned by the study sponsor, the HPS Network. The HPS Network in collaboration with the National Organization for Albinism and Hypopigmentation (NOAH) 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.

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

In all cases, your privacy will be protected. The Registry Advisory Board 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 to keep your information safe.

26. How is the registry maintained?

The registry is maintained by NORD who hosts the registry on its web-based application. NORD provides ongoing technical support of the system. The HPS Network provides the day-to-day management of their patient registry.

27. 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/.