REFSUM DISEASE
PATIENT REGISTRY
Presenters

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Sanford Research

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AGENDA

Introduction to Global DARE Foundation
Patient Registry Overview
Introduction to Sanford CoRDS
Registry Demonstration
Question & Answer Session
WEBINAR HOUSEKEEPING DETAILS

- All participants are in listen only mode
- How to ask a question during the Q&A:
  - Participants following on Zoom can type their questions in the Q&A box at any time during the presentation or by raising their hand at the end to ask a question live.
  - Participants joining by phone can press *9 on their phone to raise their hand.
- Questions will be answered in the following order:
  - Q&A box in Zoom
  - Dial in participants
  - Online participants
- Today’s session will be recorded for later viewing on Global DARE Foundation Website (www.defeatadulttrefsumeverywhere.com)
DARE’S MISSION

Global DARE Foundation's mission is to promote world-wide awareness and better quality of life for all who are diagnosed with Adult Refsum Disease.
WHY A PATIENT REGISTRY?

Simply PROGRESS!

A patient registry will bring the Refsum community and researchers together to gain a better understanding of Refsum Disease. Global DARE Foundation’s registry at Sanford CoRDS will help to do the following:

- Help develop treatment guidelines to improve the care people receive
- Drive specific research to develop better therapies
- Provide Global DARE Foundation information to focus support efforts
- Represent a link between Researchers and the Refsum community
- Create interest from investors that can help fund therapy development
- Identify patients for clinical trial

We need support from the whole community to make this happen.
REGISTRY HIGHLIGHTS

• Each participant is assigned a Global Unique Identifier that enables de-identification of the data when shared with approved researchers to protect the patient's privacy. This means that your name and other identifying information will not be passed on.

• The de-identified data will be shared only with researchers approved by the Global DARE Foundation and Sanford's Scientific Advisory Board (SAB).

• The registry is compliant with the European Union General Data Protection Regulation (GDPR).

• Importantly, the participant owns his/her personal data and can withdraw the data from the registry at any time.

• There is no cost to the participants.

• Providing your consent to Global DARE Foundation to have access to the data you provide will allow us to better understand Refsum disease, help us understand where to drive our research initiatives, and let us know how patients need more support.
Some examples of the types of data you will be asked to enter in the registry include:

- Clinical Diagnosis
- Current Symptoms
- Ongoing Refsum Management
- Phytanic Acid levels
- Dietary Information
- Quality of Life Information
- General Health Information
What is CoRDS?

• CoRDS is a patient registry for all rare diseases, unaffected carriers & the undiagnosed - it ties together patients, advocacy groups, and researchers.

• CoRDS is made available at no cost to patients, advocacy groups or researchers. It is free and it always will be.
Who can Access my Data?

**Researchers:** The de-identified information in CoRDS will be made available to researchers studying rare, uncommon or undiagnosed disorders.

**Other Patient Registries:** A subset of de-identified information collected from each profile can be shared with some other databases.

**Patient Advocacy Groups:** Patient advocacy groups (PAGs) representing individuals with rare or uncommon diseases who have partnered with CoRDS may also request access to information in CoRDS that may or may not include participants’ names.
Is my Data Safe?

• Yes, we take your privacy and security seriously. Your identifiable information can only be accessed by CoRDS personnel and any patient advocacy group with whom you consent to share your information.
CoRDS is a centralized international patient database for all rare diseases. Our goal is to connect as many patients and researchers as possible to advance treatments and cures for rare diseases.
How to Enroll

- Participants will first go to Global DARE Foundation website at the following link: https://www.defeatadultrefsumeverywhere.org/refsum-patient-registry

- The website has links to the CoRDS Refsum registry, tips for navigating the registry platform as well as answers to frequently asked questions.
Activation Form

Introduction
Please answer a few questions to help us create your participant account. If you have any questions, please contact cords@sanfordhealth.org or 1 (877) 658-9192.

Participant Type
- I am enrolling myself (18 years or older)
- I am enrolling my child (child under the age of 18)
- I am enrolling an adult who is not cognitively able to enroll (Must be the Legally Authorized Representative (LAR))

Participant Information
Please provide your basic information. Red asterisk (*) indicates a required field.

Participant First Name *
Participant Middle Name
Participant Last Name *

Birth Date *

Do you speak and understand English? *
- Yes
- No

Please Select a language

If you selected “Other”, please specify

Enrollment Information
## Diagnosis

Please select the participant's diagnosis from the "Rare Disease Diagnosis" section. You can add or remove multiple diagnosis from the field with a maximum entry of 5 diseases.

- If you have not officially been diagnosed with a Rare Disease, please enter "Undiagnosed or Unaffected Carrier" (along with a rare disease) if applicable to the participant.
- If you cannot find your disease search for "Other" and add your diagnosis in the Other Rare Diseases Diagnosis below.

<table>
<thead>
<tr>
<th>Rare Disease Diagnosis 1</th>
<th>Refsum disease</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rare Disease Diagnosis 2</td>
<td></td>
</tr>
<tr>
<td>Rare Disease Diagnosis 3</td>
<td></td>
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<tr>
<td>Rare Disease Diagnosis 4</td>
<td></td>
</tr>
<tr>
<td>Rare Disease Diagnosis 5</td>
<td></td>
</tr>
</tbody>
</table>
| Specify Other Rare Disease Diagnosis | }
Activation Form

Consent

There are no direct benefits to participating in CoRDS but the data you contribute may help advance research for rare diseases which would benefit a wider population. By participating in CoRDS you may be contacted about research opportunities you qualify for, and can decline that time if you would like to participate, however CoRDS cannot guarantee that a researcher will request to contact you.

Is the information kept confidential? What are the risks of participation?
CoRDS will make every effort to keep all information gathered in the registry confidential, but this cannot be guaranteed. There is a minimal risk associated with the loss of confidentiality.

How will my information be accessed? Can I choose how my information is shared?
On the questionnaire(s), you can choose how you would like CoRDS to share your Information. Information in CoRDS may be accessed in the following ways:

1. Researchers may access de-identified information once they are approved by the CoRDS Scientific Review Committee. If you enter a disease-specific registry for a Patient Advocacy Group (PAG), that PAG may ask to be involved in the review process. If a researcher wishes to notify you about a research opportunity, CoRDS Personnel will contact you on their behalf. There is no obligation to participate - it is always up to you to contact the researcher. CoRDS ensures that all research study referrals have been reviewed and approved by an IRB as applicable.
2. A subset of de-identified information may be shared with certain other databases in order to avoid a duplication of efforts and to increase knowledge and understanding of rare diseases.
3. Patient Advocacy Groups (PAGs) representing individuals with rare or uncommon diseases may have access to information that may or may not be identifiable for non-research purposes. These PAGs have signed a contract stating they will not use the information for research purposes.

Who should I contact if I have questions?

- For general questions about CoRDS or enrollment: call (877) 658-9192 or email cords@sanfordhealth.org
- If you feel you have been harmed: Benjamin Forred, (605) 312-6416 if you have questions about your rights as a research participant: Sanford Health Institutional Review Board (IRB), (605) 312-6430

By checking this box, I agree to participate in the CoRDS Registry

Submit
Login Screen

Login Setup

Username: Polly.gill4
New Password: ********
Confirm New Password: ********
Security Question: Select an Option
Security Answer:

Login
Demonstration of the CoRDS Registry
# REFSUM SPECIFIC QUESTIONS

<table>
<thead>
<tr>
<th>Question Category</th>
<th>Data Collected</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Permission and Data Sharing</strong></td>
<td>Checking Global DARE Foundation will allow your data to be shared with the foundation to help drive research, build targeted programs and help our community to better understand the disease.</td>
</tr>
</tbody>
</table>
| **At Diagnosis**               | - Symptoms at diagnosis including specifics around vision, hearing and phytanic acid level  
    - Diagnostic tests conducted to diagnosis as well as confirm diagnosis  
    - Asks for when Retinitis Pigmentosa was diagnosed to understand the length of time between the RP diagnosis and the Refsum diagnosis                                                                                                                                 |
| **Ongoing Refsum Management**  | - Current symptoms and conditions  
    - Specialists seen and diagnostic tests conducted regularly  
    - Current and historical phytanic acid levels                                                                                                                                                                                                                                  |
| **Plasmapheresis**             | Frequency of plasmapheresis and the triggers for when it is done                                                                                                                                                                                                                                                                                  |
| **Low Phytanic Acid Diet**     | Use of a low phytanic acid diet and general eating routines                                                                                                                                                                                                                                                                                     |
| **Quality of Life**            | How Refsum Disease may impact quality of life                                                                                                                                                                                                                                                                                                     |
| **Mobility**                   | How Refsum Disease may impact mobility                                                                                                                                                                                                                                                                                                            |
| **Exercise**                   | Exercise routines including any limitations due to Refsum Disease                                                                                                                                                                                                                                                                                  |
| **General Health / Other Info**| General health information (e.g. weight, height) as well as other medical conditions, surgeries or medicine/supplements that may apply to a patient.                                                                                                                                                                                                     |
| **Pregnancy Information**      | Data on female patients that have had children                                                                                                                                                                                                                                                                                                    |
**IMPORTANT TIPS TO REMEMBER**

- **Screen Reader:** The CoRDS registry is currently not compatible with a screen reader. Therefore, assistance may be necessary if you require the use of a screen reader.

- **Device:** You should use a computer to complete the survey and not a tablet or smart phone.

- **Zoom:** It may be necessary to modify the Zoom settings on your screen to have the best visual experience when completing the questionnaire.

- **Saving:** You can save the questionnaire at any point, close out and log back into the registry later. Even if you have clicked FINISH at the end of the questionnaire, you can go back at any time and update your answers.

- **Difficult questions:** If you encounter difficult questions, please feel free to move on to other questions and come back to more difficult ones later.

- **Answering questions:** Some find it easier to go through the entire questionnaire answering “easy” questions first and noting which questions they need more time to answer.

- **Paper Version:** Completing the questionnaire in paper form is possible and only needs to be requested from Sanford CoRDS.