Questionnaires

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CoRDS Registry

Coordination of Rare Diseases at Sanford

Instructions

Thank you for taking the time to enroll with the CoRDS Registry. This questionnaire:

- Takes 10 - 20 minutes to complete
- Will refer to the person with the rare or unknown diagnosis as "**the participant**"
- Can be updated at any time by logging in to the CoRDS online portal or by contacting CoRDS personnel

CoRDS personnel will contact you annually to update your questionnaire.

If you have any questions while completing this form, please contact CoRDS at (877) 658 – 9192 during business hours, 8:30am-5:00pm (CST) Monday through Friday. If you need assistance after business hours, please leave a message or email cords@sanfordhealth.org.

*For accurate data curation, please remember to write legibly. Thank you.

1. Today's Date (MM/DD/YYYY):

2. Who is completing this questionnaire?

☐ I am enrolling myself (You must be over the age of 18 to provide information for the registry)

☐ I am enrolling my child (You must be the participant's parent or legal guardian to provide information for the registry)

☐ I am enrolling an adult who is not cognitively able to enroll (You must be the participant's legally authorized representative (LAR) to provide information for the registry)

Permissions & Data Sharing

By participating in CoRDS, your de-identified information will be shared with researchers who access the CoRDS Registry. Below are options that allow you to share your data with other entities. In the following questions, please select how you want your data shared. Please complete this section before moving on.

3. I give permission to CoRDS to contact me about participating in future research studies:

☐ Yes ☐ No ☐ Don't Know

4. I give permission to CoRDS to contact me about donating a sample of blood, tissue, or other biospecimen for research in the future:

☐ Yes ☐ No ☐ Don't Know

5. I give permission to CoRDS to provide a subset of de-identified information to other databases collecting information on rare diseases in order to avoid a duplication of efforts and to increase knowledge:

☐ Yes ☐ No

Participant Information

6. First Name:	**7. Middle Name:**	**8. Last Name:**
☐ Check if the legal given name (as per birth certificate) of the participant is the same as indicated above		
Legal given name of the participant (as per birth certificate)		
9. First Name:	**10. Middle Name:**	**11. Last Name:**
12. Date of Birth:	**13. City, Town, or Village of Birth:**	**14. Country of Birth:**
15. Current Address 1:		
16. Current Address 2:		
17. Current City, Town, or Village:	**18. Current State or Province:**	

19. Zip/Postal Code	**20. Country**
21. Email Address:	
22. Primary Telephone Number:	

Parent / Legally Authorized Representative (LAR) Information

Please complete this section if you are the participant's parent/guardian (**participant must be under the age of 18**) or legally authorized representative (**participant is not cognitively able to enroll).**

23. First Name:	
24. Middle Name:	
25. Last Name:	
26. Primary Telephone Number:	
27. Email Address:	
☐ Check if the address is the same as the participant's, then skip to next section	
28. Address 1:	
29. Address 2:	
30. City, Town or Village	**31. State or Province**
32. Zip/Postal Code:	**33. Country:**

Secondary Contact

Please provide information for an individual that we may contact in the event that we are unable to reach you.

34. Relationship to Secondary Contact:

☐ Family Member ☐ Spouse/Partner
☐ Friend ☐ Other

35. If you selected "Other" above, please specify: _____

36. First Name:	
37. Middle Name:	
38. Last Name:	
39. Primary Telephone Number:	
40. Email Address:	
☐ Check if the address is the same as the participant's, then skip to next section	
41. Address 1:	
42. Address 2:	
43. City, Town or Village:	
44. State or Province:	
45. Zip/Postal Code:	**46. Country:**

Enrollment, Contact & Communication Preferences

47. Special Communication Needs: Do you (the person completing this form) have any special communication needs? Please select all that apply, or describe in the space provided.

☐ No special needs - both spoken and written language are acceptable
☐ Sign language required
☐ Spoken language preferred
☐ Written language preferred
☐ Other

48. If you selected "Other" above, please specify: _____

Participant Socio-demographic Information

Please provide information about the participant's background and diagnosis in the following sections.

49. Sex:	☐ Female	☐ Male	☐ Transsexual	☐ Unknown	☐ Other
50. Sex at Birth:	☐ Female	☐ Male	☐ Transsexual	☐ Unknown	☐ Other

51. Race:

☐ American Indian or Alaska Native
☐ Asian - Asian Indian
☐ Asian - Chinese
☐ Asian - Filipino
☐ Asian - Japanese
☐ Asian - Korean
☐ Asian - Vietnamese
☐ Asian - Other Asian
☐ Black or African American
☐ Pacific Islander – Native Hawaiian
☐ Pacific Islander - Guamanian
☐ Pacific Islander - Chamorro
☐ Pacific Islander- Samoan
☐ Pacific Islander - Other Pacific Islander
☐ White
☐ Other/Unknown/Refuse to Answer

52. Ethnicity:

☐ Not Hispanic or Latino
☐ Ashkenazi Jewish
☐ French Canadian
☐ Hispanic or Latino - Central American
☐ Hispanic or Latino - Cuban
☐ Hispanic or Latino - Dominican (Republic)
☐ Hispanic or Latino - Mexican
☐ Hispanic or Latino - Puerto Rican
☐ Hispanic or Latino - South American
☐ Hispanic or Latino - Other Latin American
☐ Hispanic or Latino - Other Hispanic/Latino/Spanish
☐ Unknown/No answer
☐ Other

53. If you selected "Other" above, please specify: _____

54. Is the participant still living?

55. If you selected "No" above, please indicate date of death (MM/DD/YYYY): _____ **Caus**

Diagnosis

56. For genetic rare diseases, is the participant an unaffected carrier of the rare disease?

☐ Yes	☐ No	☐ Unknown

57. If you selected "Yes" above, please list the rare disease for which the participant is a carrier for.

58. Rare Disease Diagnosis: Please list all rare disease diagnoses.

Please complete the questions below in relation to the disease identified above. If you have more than one rare disease, please answer questions 58 and 61-68 for each condition.

59. Rare Disease Symptoms: Please list symptoms of rare disease diagnosis. Separate with commas.

60. Undiagnosed: If no clinical diagnosis has been made, please list symptoms. Separate with commas.

61. Other Diagnoses: Please list non-rare diagnoses. Separate with commas.

62. Age at Diagnosis: ☐ Prenatal ☐ At birth ☐ Age ☐ Unknown ☐ N/A

63. If you selected "Age" above, please indicate age: (years and/or months) _____

64. Age at First Symptom: ☐ Prenatal ☐ At birth ☐ Age ☐ Unknown ☐ N/A

65. If you selected "Age" above, please indicate age: (years and/or months) _____

66. How was the rare diagnosis determined? Select all that apply.

☐ Genetic Laboratory Analysis
☐ Histology
☐ Imaging – CT
☐ Imaging – MRI
☐ Imaging – PET
☐ Physical Examination
☐ Unknown
☐ Other

67. If you selected "Other" above, please specify:

68. Where was the diagnosis made?

Hospital / Institution: _____

City: _____

State or Province: _____

Country: _____

Family History

69. Which family members also have the participant's rare disease? Select all that apply.

☐ None	☐ Unknown
☐ Mother	☐ Paternal Grandmother
☐ Father	☐ Maternal Aunt
☐ Brother	☐ Paternal Aunt
☐ Half-brother	☐ Maternal Uncle
☐ Sister	☐ Paternal Uncle
☐ Half-sister	☐ Maternal Cousin
☐ Daughter	☐ Paternal Cousin
☐ Son	☐ Granddaughter
☐ Maternal Grandfather	☐ Grandson
☐ Paternal Grandfather	☐ Niece
☐ Maternal Grandmother	☐ Nephew

Quality of Life

70. In general, would the participant say his/her health is…

☐ Excellent	☐ Very good	☐ Good	☐ Fair	☐ Poor

71. Does the participant's health now limit him/her in doing vigorous activities?

☐ Never	☐ Rarely	☐ Sometimes	☐ Often	☐ Always

72. How much did pain interfere with the participant's enjoyment of life?

☐ Never	☐ Rarely	☐ Sometimes	☐ Often	☐ Always

73. How often does the participant feel tired?

☐ Never	☐ Rarely	☐ Sometimes	☐ Often	☐ Always

74. The participant feels depressed…

☐ Never	☐ Rarely	☐ Sometimes	☐ Often	☐ Always

Clinical Research Participation & Biospecimens

75. Has the participant *previously participated in any clinical trials* related to their rare disease?

☐ Yes ☐ No ☐ Don't know

76. Does the participant *currently participate in any clinical trials* related to their rare disease?

☐ Yes ☐ No ☐ Don't know

77. Has the participant *previously donated a sample* of blood, tissue, or other biospecimen for research?

☐ Yes ☐ No ☐ Don't know

78. If Yes:

Type of biospecimen:

☐ Blood ☐ Tissue

☐ Other bodily fluid ☐ Urine

☐ Saliva/Cheek Swab ☐ Unknown

79. Location of biospecimen donation:

☐ Check here if location unknown

Hospital/ Institution:

City: _____

State or Province: _____

Country: _____

Thank you for your participation!

Questions?

CoRDS Personnel

Sanford Research

2301 East 60th Street North

Sioux Falls, South Dakota 57104

Phone (toll-free)**:** 1 (877) 658-9192

Email: CoRDS@sanfordhealth.org

Instructions

Thank you for taking the time to enroll with the CoRDS Registry. This module will ask you questions specific to your diagnosis. The questions below were developed in partnership with Global DARE Foundation. Please note, this module:

- Takes approximately 30 minutes to complete
- Will refer to the person with the diagnosis as "**the participant**"
- Can be updated at any time by logging in to the CoRDS online portal or by contacting CoRDS personnel

If you have any questions while completing this form, please contact CoRDS at (877) 658-9192 during business hours, 8:30am-5:00pm (CST) Monday through Friday. If you need assistance after business hours, please leave a message or email cords@sanfordhealth.org.

Permissions & Data Sharing

I give permission to CoRDS to provide my information that may or may not be identifiable to the following Patient Advocacy Group (PAG) for non-research purposes.

☐ Global DARE Foundation	☐ I do not give my permission

At Diagnosis

1. What symptoms were present at the time of the participant's Refsum diagnosis? (select all that apply):

☐ Anosmia (loss of smell)	☐ Ichthyosis (scaly, dry skin)
☐ Ataxia (unsteadiness, balance issues)	☐ Peripheral neuropathy (nerve pain in limbs, numbness)
☐ Other bone abnormalities (joint issues)	☐ Retinitis pigmentosa (loss of vision)
☐ Cardiac signs	☐ Shortened metacarpals and metatarsals (shorten toes or fingers/thumbs)
☐ Hearing impairment	☐ Other

If "other", please specify:

2. If the participant has Retinitis Pigmentosa, at what age did the participant receive their Retinitis Pigmentosa diagnosis?_____years

3. What was the participant's degree of peripheral vision when diagnosed with Refsum through a visual field test? _____

4. What was the participant's degree of hearing loss at diagnosis?

☐ None	☐ 51% - 75%
☐ < 25%	☐ > 75%
☐ 25% - 50%	☐ No hearing

5. What medical tests were performed at diagnosis?	
☐ Brain MRI	☐ Optical coherence tomography
☐ Echocardiogram	☐ Phytanic Acid level
☐ Electroretinography (ERG)	☐ Visual field test
☐ Hearing test	☐ X-Rays
☐ Nerve conduction	☐ Other
If "other", please specify (separate by commas if more than one):	
6. What was the participant's Phytanic Acid level at diagnosis? (Please use measurement pertaining to the participant's lab test) _____µmol/L or_____mg/dL	
7. What is the participant's gene mutation?	
☐ PHYH	☐ Other
☐ PEX7	☐ Unknown
If "other", please specify:	
8. Was a fibroblast test (skin biopsy) conducted?	
☐ Yes	☐ No
If "yes", what were the results?	
Ongoing Refsum Management	
9. What are the participant's current symptoms? (select all that apply):	
☐ Anosmia (loss of smell)	☐ Ichthyosis (scaly, dry skin)
☐ Ataxia (unsteadiness, balance issues)	☐ Peripheral neuropathy (nerve pain in limbs, numbness)
☐ Other bone abnormalities (joint issues)	☐ Retinitis pigmentosa (loss of vision)
☐ Cardiac signs	☐ Shortened metacarpals and metatarsals (shorten toes or fingers/thumbs)
☐ Hearing impairment	☐ Other
If "other", please specify:	
10. Does the participant use a hearing device?	
☐ Yes	☐ No
11. If "yes", what type of hearing device does the participant use?	

☐ Cochlear implants	☐ Other	
☐ Hearing aids		
If "other", please specify:		
12. What is the participant's current degree of peripheral vision through a visual field test? _____		
13. Did the participant have cataract surgery?		
☐ Yes	☐ No	
14. Is the participant being treated for macular edema?		
☐ Yes	☐ No	
15. Does the participant's vision require the use of any of the following? (select all that apply):		
☐ Guide dog	☐ Signal cane	
☐ Long cane	☐ None	
16. How many Refsum related hospital admissions has the participant had?		
☐ None	☐ < 5 since diagnosis	
☐ Only at diagnosis	☐ > 5 since diagnosis	
17. How often does the participant see the following specialists?		
Audiologist	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Cardiologist	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Dietician	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Ear, Nose, & Throat Doctor (ENT)	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Metabolic Specialist	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never

Neurologist	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Ophthalmologist	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Other: _____	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Other: _____	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
18. How frequently are the following medical tests performed?		
Brain MRI	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Darkness adaptation test	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Echocardiogram	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Electroretinography (ERG)	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Hearing test	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Nerve conduction test	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never

Optical coherence tomography (OTC)	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Visual field test	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Other: _____	☐ More frequently than yearly ☐ Yearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never
Other: _____	☐ More frequently than yearlyYearly ☐ Every 2 years ☐ Every 3 years	☐ Less frequently than every 3 years ☐ Only at diagnosis ☐ Never

19. What is the participant's typical daily pain level?

☐ None	☐ Moderate (4 to 6)
☐ Low (1 to 3)	☐ High (7 to 10)

20. How often does the participant have a Phytanic Acid Level done?

☐ Every month	☐ Every 6 months
☐ Every other month	☐ Yearly
☐ Every 3 months	☐ Other

If "other", please specify:

21. When was the participant's last PhytanicAcid level?_____(DD/MM/YYYY)

22. What was the participant's last Phytanic Acid level? (Please use measurement pertaining to the participant's lab test) _____µmol/L or_____mg/dL

23. What was the participant's highest Phytanic Acid level since diagnosis? (Please use measurement pertaining to the participant's lab test) _____µmol/L or_____mg/dL

24. What was the participant's lowest Phytanic Acid level since diagnosis? (Please use measurement pertaining to the participant's lab test) _____µmol/L or_____mg/dL

25. What was the participant's highest Phytanic Acid level in the last 3 years? (Please use measurement pertaining to the participant's lab test _____µmol/L or _____mg/dL

26. What was the participant's lowest Phytanic Acid level in the last 3 years? (Please use measurement pertaining to the participant's lab test) _____µmol/L or _____mg/dL

27. What is the participant's average Phytanic Acid level over the last 3 years?	
☐ Under 100 **µmol/L** (under 3.13 **mg/dl**)	☐ 301 – 500 **µmol/L** (9.39 – 15.63 **mg/dl**)
☐ 100 – 300 **µmol/L** (3.13 - 9.38 **mg/dl**)	☐ Above 500 **µmol/L** (15.63 **mg/dl**)
Plasmapheresis	
28. How many times has the participant had plasmapheresis?	
☐ Never	☐ 5 – 10 times
☐ 1 – 4 times	☐ > 10 times
29. If the participant has had plasmapheresis, what is the indication/trigger for it to be done? (select all that apply):	
☐ Acute symptoms	☐ Routinely done
☐ High levels of Phytanic Acid	
30. If the participant has plasmapheresis routinely, how often is it done?	
☐ Monthly	☐ Every 6 months
☐ Every 6 weeks	☐ Yearly
☐ Every 2 months	☐ Other
☐ Every 3 months	
If "other", please specify:	
Low Phytanic Acid Diet	
31. Is the participant on a Low Phytanic Acid diet?	
☐ Yes	☐ No
32. What is the participant's compliance with the low phytanic acid diet within the last year?	
☐ Very strict	☐ Poor
☐ Good	☐ Not following the diet

☐ Intermittent	
33. How many times a day does the participant eat including snacks?	
☐ < 3	☐ > 6
☐ 3 - 6	
34. On average, what is the longest period during the day the participant would go without eating between meals/snacks?	
☐ < 2 hours	☐ 4 – 6 hours
☐ 2 – 3 hours	☐ > 6 hours
35. What impact does the low phytanic acid diet have on the participant's quality of life? (e.g. eating out, social activities)	
☐ No impact	☐ Moderate impact
☐ Low impact	☐ High impact
36. Is the participant's Phytanic Acid level or disability relating to Refsum a barrier to maintaining the participant's desired weight?	
☐ Yes	☐ No
Quality of Life	
37. Is the participant discouraged about having Refsum Disease?	
☐ None of the time	☐ A good bit of the time
☐ A little bit of the time	☐ Most of the time
☐ Some of the time	☐ All of the time
38. Is the participant worried about having Refsum Disease?	
☐ None of the time	☐ A good bit of the time
☐ A little bit of the time	☐ Most of the time
☐ Some of the time	☐ All of the time
39. Which of the following categories best describes the participant's employment status?	

☐ Student	☐ Not employed, looking for work
☐ Employed, part-time	☐ Not employed, not looking for work
☐ Employed, full-time	☐ Retired
☐ Stay at home parent	☐ Disabled, not able to work
40. If not working or working part-time, is this due to Refsum Disease?	
☐ Yes	☐ No
41. Has a participant changed jobs due to Refsum Disease?	
☐ Yes	☐ No

Mobility

42. What is the participant's level of mobility?
☐ No mobility
☐ Limited (requires full assistance)
☐ Moderate (walk semi-assisted, need a railing to walk up and down stairs)
☐ High (can walk unassisted, very steady, able to walk on uneven surfaces)
43. How would the participant describe the current level of balance?

☐ Good	☐ Poor
☐ Moderate	

44. What devices (if any) does the participant use for mobility? (separate by commas):

Exercise	
45. How many days in a week does the participant usually exercise?	
☐ 0	☐ 4 – 6
☐ 1	☐ 7
☐ 2 - 3	
46. What type(s) of activity/exercise does the participant engage in? (select all that apply):	
☐ Bowling	☐ Mobility exercises (yoga, stretching, Pilates)
☐ Cricket	☐ Swimming
☐ Curling	☐ Walking
☐ Cycling	☐ Yard/housework
☐ Jogging	☐ Not applicable
☐ Lifting weights	☐ Other
If "other", please specify:	
47. Over the course of one week, what is the total number of minutes the participant exercise or does an activity?	
☐ Not active/does not exercise	☐ 91 – 120 minutes
☐ <30 minutes	☐ 121 – 150 minutes
☐ 31 – 60 minutes	☐ > 150 minutes
☐ 61 – 90 minutes	
48. What is the participant's maximum exercise intensity level?	
☐ No exercise	☐ Moderate (cycling, swimming)
☐ Light (yoga, walking)	☐ Hard (running, interval training)

49. Has Refsum Disease prevented the participant from doing certain type(s) of exercise?

☐ Yes	☐ No

If "yes", please specify:

General Health / Other Information

50. What is the participant's current weight? _____lbs or _____kg

51. What is the participant's current height? _____inches or_____centimeters

52. Does the participant have any other medical conditions? (select all that apply):

☐ Arthritis	☐ Heart disease	☐ Obesity
☐ Anxiety	☐ High blood pressure	☐ Pre-diabetes
☐ Asthma	☐ High cholesterol	☐ Previous stroke
☐ Cancer	☐ Liver disease	☐ None
☐ Depression	☐ Low back pain	☐ Other
☐ Diabetes	☐ Kidney disease	

If "other", please specify:

53. Has the participant ever had problems with sweating?

☐ Yes	☐ No

If "yes", please explain:

54. Has the participant had any surgeries?

☐ Yes	☐ No

If "yes", please explain. (separate by commas):

55. Does the participant often have cold hands or feet?

☐ Yes	☐ No
56. Does the participant have joint pain in the any of the following? (select all that apply):	
☐ Ankle	☐ Shoulder
☐ Elbow	☐ Wrist
☐ Hip	☐ No joint pain
☐ Knee	☐ Other
If "other", please specify:	
57. What medications (prescription and over the counter) does the participant currently take? (separate by commas):	
58. What vitamin supplements does the participant take? (separate by commas):	
Pregnancy (if applicable)	
59. If female, did the participant have children after diagnosis of Refsum Disorder?	
☐ Yes	☐ No
60. If "yes", were there any complications?	
☐ Yes	☐ No
61. Was the participant's Phytanic Acid levels monitored during pregnancy?	
☐ Yes	☐ No
62. Did the participant breast-feed?	
☐ Yes	☐ No
Additional Information	

63. Please provide any additional details about the participant's diagnosis that is worth noting.