Instructions

Thank you for taking the time to enroll with the Coordination of Rare Diseases at Sanford (CoRDS) Registry. This module will ask you questions specific to your diagnosis. The questions below were developed in partnership with Maple Syrup Urine Disease Family Support Group (MSUD FSG). Please note, this module:

- Takes approximately 40 minutes to complete – does not need to be completed in one sitting
- Can be updated at any time by logging in to the CoRDS online portal or by contacting CoRDS personnel
- Will refer to the person with the diagnosis as "**the participant**"

We gratefully acknowledge the work of the Emory University Metabolic Genetics team in developing the NBS Connect registry with a grant from HRSA, which served as a template for this survey.

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.

***By granting MSUD Family Support Group permission to receive data, this means MSUD Family Support Group will receive a report of all de-identified data on a weekly basis. De-identified information is information that cannot identify an individual. The MSUD Family Support Group may request the participant's name and contact information so they can contact the participant with information about the support group. This information will be provided as a separate report and not associated with the participant's responses to any other questions.*

☐ MSUD Family Support Group	☐ I do not give my permission

Diagnosis

1. What type of MSUD has the participant been diagnosed with?

☐ Classic	☐ Other
☐ Intermittent	☐ Unknown
☐ Intermediate	

If "other", please specify:	
2. What was the participant's blood leucine level at diagnosis? (Please answer in Mg/dL OR µmol/liter):	
_____Mg/dL_____µmol/liter	☐ Unknown
3. What were the symptoms of the participant upon diagnosis? (select all that apply):	
☐ Ataxia (loss of control of movements)	☐ Nausea/vomiting
☐ Coma	☐ Poor concentration
☐ Delayed developmental milestones	☐ Poor feeding
☐ Dystonia (involuntary muscle contractions)	☐ Seizures
☐ High pitched cry	☐ Skin rash
☐ Hypertonia (increase in muscle tension)	☐ Weakness
☐ Hypotonia (reduced muscle strength)	☐ Weight loss
☐ Irritability	☐ Other
☐ Lethargy	☐ None
☐ Maple syrup odor (urine, ear wax)	
If "other", please specify:	
4. Has the participant had genetic testing?	
☐ Yes	☐ No
5. Which gene is affected (the participant's clinic may be able to provide this information):	
☐ BCKDHA	☐ DLD
☐ BCKDHB	☐ Don't know
☐ DBT	
6. What is the specific variant(s) identified as the cause of MSUD:	
☐ Y438N (c.1312T>A (p.Tyr438 Asp) Common Mennonite Variant	☐ IVS3[+3]delA
☐ R183P (c.548G>C (p.Arg183Pro) Common Ashkenazi Variant	☐ IVS5-1G > C
☐ R242X (c.859C>T (p.Arg242Ter)	☐ Don't know
☐ c.662_663delCC	☐ Other

If "other", please specify:		
7. Did the participant's biological parents have pre-natal genetic testing?		
☐ Yes	☐ No	☐ Unsure
8. Did the participant's biological parents know they were at high risk for having a child with MSUD?		
☐ Yes	☐ No	☐ Not applicable

Medical History

9. How many times has the participant been hospitalized in the past 12 months?

☐ 0	☐ 3 – 4
☐ 1 – 2	☐ 5 or more

10. Did the participant require MSUD TPN during any of the hospitalizations in the past 12 months?

☐ Yes	☐ No	☐ Was not hospitalized in the past 12 months

If "yes", how many times did the participant receive MSUD TPN? _____

11. Did the participant require dialysis during any of the hospitalizations in the past 12 months?

☐ Yes	☐ No

If "yes", how many times did the participant receive dialysis? _____

12. How many times has the participant been hospitalized in their lifetime?

☐ 0	☐ 5 – 9
☐ 1 – 4	☐ 10 or more

13. How many times has the participant received MSUD TPN in their lifetime?

☐ 0	☐ 3 – 4
☐ 1 – 2	☐ 5 or more

14. How often is the participant currently seen by a metabolic specialist for routine check-ups?

☐ Weekly/biweekly	☐ Annually
☐ Monthly	☐ Less than once a year
☐ Quarterly (every 4 months)	☐ Does not follow a metabolic specialist at this time
☐ Every 6 months	

15. Which metabolic clinic is the participant treated at?

Clinic Name: _____

City: _____

State:_____

Country if not in the United States:_____

16. Other than when ill, how often are the participant's blood amino acids measured?

☐ Twice Weekly	☐ Every 6 months
☐ Weekly	☐ Annually
☐ Monthly	☐ Less than once a year
☐ Quarterly (every 4 months)	☐ Other

If "other", please specify:

17. How long does it take for the participant to receive blood amino acid results after routine check-ups?

☐ Within 24 hours	☐ 5 – 7 days
☐ 24 – 48 hours	☐ > 1 week
☐ 3 – 4 days	

18. Does the participant currently measure urinary ketones?

☐ Yes	☐ No

If "yes", how frequently does the participant measure urinary ketones?

☐ Daily	☐ Annually
☐ Weekly	☐ Only when ill
☐ Monthly	☐ Other

19. Does the participant have other known health conditions? (select all that apply):

☐ Celiac disease	☐ Osteopenia/osteoporosis
☐ Diabetes	☐ Seizure disorder
☐ High cholesterol/high triglycerides	☐ Short stature
☐ Hypertension/high blood pressure	☐ Thyroid Disease

☐ Movement disorder/spasticity	☐ Other

☐ Obesity	
If "other", please specify other health conditions:	
20. What is the participant's current height? (Please answer in feet and inches OR centimeters): **_____feet_____inches _____centimeters**	
21. What is the participant's current weight? (Please answer in pounds OR kilograms): **_____pounds_____kilograms**	
22. Has the participant ever been diagnosed with any of the following disorders? (select all that apply):	
☐ Anxiety Disorder	☐ Panic Disorder
☐ Attention Deficit Hyperactivity Disorder (ADHD)	☐ Personality Disorder
☐ Attention Deficit Disorder (ADD)	☐ Post-Traumatic Stress Disorder
☐ Autism	☐ Schizophrenia
☐ Bipolar Disorder	☐ Social Phobia
☐ Eating Disorder	☐ Specific Phobia
☐ Intellectual impairment	☐ Other
☐ Major Depressive Disorder	☐ None
☐ Mood Disorder	☐ Unsure
☐ Obsessive Compulsive Disorder	
If "other", please specify:	
23. Has the participant ever been treated by any of the following? (select all that apply):	

	Currently being treated	**Previously been treated**	**Never been treated**
Behavior therapist	☐	☐	☐
Feeding therapist	☐	☐	☐
Occupational therapist	☐	☐	☐
Physical therapist	☐	☐	☐
Speech therapist	☐	☐	☐
Other	☐	☐	☐

24. If the participant has been diagnosed with ADD or ADHD, does the participant take medication for this condition?		
☐ Yes	☐ No	☐ Not applicable
25. If the participant has been diagnosed with depression, anxiety, or any other emotional disorder, does the participant take medication for any of these conditions?		

☐ Yes	☐ No	☐ Not applicable

26. Has the participant experienced a pregnancy?

☐ Yes	☐ Not applicable
☐ No	

If "yes", how many pregnancies has the participant experienced?

Liver Transplant

27. Which of the following best describes the participant's status with a liver transplant?

☐ Participant has already undergone a liver transplant	☐ Participant decided against liver transplant
☐ Participant is on the wait list awaiting a liver transplant	☐ Participant has not explored liver transplant
☐ Participant is considering/exploring liver transplant	

28. How old was the participant at the time of the liver transplant?_____ Years

29. If the participant had a liver transplant, were there surgical complications?

☐ Yes	☐ No

If "yes", what type of surgical complications did the participant have? (please separate by commas):

30. Does the participant take any immune-suppressive medications?

☐ Yes	☐ No

If the participant takes immune-suppressive medications and has experienced a major infection or malignant complication related to the immune-suppressive medication, please list all infections or complications:

31. If the participant takes immune-suppressive medications, please list them (separate by commas):

32. If the participant has undergone a liver transplant, which of the following best describes the donor?

☐ Living related donor	☐ Deceased donor
☐ Living, non-related donor	

33. Which of the following best describes the type of liver transplant the participant received?

☐ Auxiliary transplant	☐ Liver segment	☐ Whole liver

Diet History

34. If the participant uses a metabolic formula, which of the following does the participant currently use? (select all that apply):

☐ Acerflex	☐ Lophlex
☐ Anamix MSUD Infant	☐ Milupa MSUD 2
☐ Anamix MSUD Junior	☐ MSUD Aid
☐ Anamix MSUD Next	☐ MSUD Cooler
☐ BCAD 1	☐ MSUD Express 15
☐ BCAD 2	☐ MSUD Gel
☐ Complex Amino Acid Bars MSD	☐ MSUD Maxamaid
☐ Complex Amino Acid Blend MSD	☐ MSUD Maxamum
☐ Complex Essential MSD	☐ Unknown
☐ Complex Junior MSD	☐ Other
☐ Ketonex – 1	☐ Not prescribed a metabolic formula
☐ Ketonex – 2	

If "other", please specify:	
35. How does the participant currently consume formula and food?	
☐ By mouth	☐ By mouth and gastrostomy tube
☐ By gastrostomy tube	☐ Other
If "other", please specify:	
36. Has the participant had a gastrostomy tube in the past?	
☐ Yes	☐ No
If "yes", what was the age of the participant when they had a gastrostomy tube? (Please answer in years OR months):_____years_____months	
How long did the participant have a gastrostomy tube in place? (Please answer in years OR months): _____years_____months	
37. Does the participant take dietary supplements/amino acid supplements?	
☐ Yes	☐ No
If "yes", please specify the dose next to the supplement. Check unknown if the participant takes the supplement but is unsure of the dose.	

Supplement	Yes/No	Dose	Dosage Unknown
Thiamine (vitamin B1)	☐ Yes ☐ No		☐ Dosage Unknown
Valine	☐ Yes ☐ No		☐ Dosage Unknown
Isoleucine	☐ Yes ☐ No		☐ Dosage Unknown
Multivitamin	☐ Yes ☐ No		☐ Dosage Unknown
Iron	☐ Yes ☐ No		☐ Dosage Unknown
Calcium	☐ Yes ☐ No		☐ Dosage Unknown
Omega-3 Fatty Acids (DHA)	☐ Yes ☐ No		☐ Dosage Unknown
Carnitine	☐ Yes ☐ No		☐ Dosage Unknown

Other: _____			☐ Dosage Unknown
Other: _____			☐ Dosage Unknown
Other: _____			☐ Dosage Unknown
Other: _____			☐ Dosage Unknown

38. Does the participant count dietary leucine?		
☐ Yes	☐ No	☐ Not applicable
If "yes", how many mg of leucine is the participant advised to include daily?_____mg		
39. Does the participant count dietary protein?		
☐ Yes	☐ No	☐ Not applicable
If "yes", how many grams of dietary protein is the participant advised to include daily?_____gm		
40. Which statement best describes how the participant adheres to their prescribed diet?		
☐ The participant consistently consumes all daily formula and strictly follows their leucine/protein prescription and diet		
☐ The person sometimes deviates from their daily formula or leucine/protein or diet by minor amounts		
☐ The person often deviates from the daily formula or leucine/protein or diet by significant amounts		
☐ The person generally does not follow a daily formula, their leucine/protein prescription, or diet		

Insurance/Cost

41. If the participant takes metabolic formula, how is the metabolic formula paid for? (select all that apply):	
☐ Medicaid/Medicare	☐ Self-pay
☐ Private insurance	☐ Other
☐ Provided by state	
If "other", please specify:	
42. If the participant takes isoleucine and valine supplements, how are the supplements paid for? (select all that apply):	

☐ Medicaid/Medicare	☐ Self-pay
☐ Private insurance	☐ Other
☐ Provided by state	
If "other", please specify:	

43. Has the participant experienced difficulties obtaining reimbursement for any of the following? (select all that apply):

☐ G tube supplies	☐ ER/Hospital visits
☐ Isoleucine/valine supplements	☐ Specially modified low protein foods
☐ Lab tests	☐ Not applicable
☐ Metabolic formula	☐ Other
If "other", please specify:	

44. What is the participant's estimated monthly out-of-pocket cost for metabolic formula and isoleucine/valine supplements? If the participant does not know the estimated monthly out of pocket cost, please leave the response blank _____

45. What is the participant's estimated total yearly out-of-pocket costs for their MSUD diagnosis? (i.e., medical visits, lab test copays, equipment supplies, etc.). If the participant does not know the estimated yearly out of pocket cost, please leave the response blank _____

Academic/Work/Social

46. If the participant is a student, does the participant require individual learning support?

☐ Yes	☐ No	☐ Not applicable

If "yes", what support does/did the participant require? (select all that apply):

☐ Aide	☐ Resource Center
☐ Extra time for assignments/exams	☐ Other
If "other", please specify:	

47. If the participant is no longer a student, did the participant require individual learning support in the past?

☐ Yes	☐ No	☐ Not applicable

If "yes", what support did they require? (select all that apply):

☐ Aide	☐ Resource Center
☐ Extra time for assignments	☐ Other

If “other”, please specify:	
48. Does the participant work in a paid position?	
☐ full time (35 - 40 hours/week)	☐ does not work in a paid position
☐ part time (20 – 35 hours/week)	☐ Not applicable
☐ less than 20 hours/week	
49. If the participant is an adult, which of the following best describes the participants living situation?	
☐ Lives alone	☐ Lives with parents

☐ Lives independently with assistance	☐ Participant is a child
☐ Lives in a group home	☐ Lives with family member other than parents
☐ Lives with spouse	☐ Other
If "other", please specify:	
50. Has the participant or their family been involved with the MSUD Family Support Group?	
☐ Yes	☐ No
If "yes", please specify which type of involvement?	
☐ Attends MSUD Family Support Group symposium	☐ Refers to MSUD Family Support Group website for information
☐ Donates to MSUD Family Support Group	☐ Follows MSUD Family Support Group Facebook Page
☐ Reads MSUD Family Support Group newsletter	☐ Other
If "other", please specify:	
50. If the participant has not been involved in the MSUD Family Support Group, is the participant or family interested in getting involved? *Please note: your name and contact information will be provided to the MSUD Family Support Group if you have given permission to MSUD FSG in the permission and data sharing section**.**	
☐ Yes	☐ No

Thank you for taking the time to respond to this survey. Your input will help shape future MSUD research and advocacy efforts. Participants are able to update this questionnaire at any time by logging into the online portal or contacting CoRDS personnel by email at cords@sanfordhealth.org or by phone 1-877-658-9192.