cystinosis RESEARCH FOUNDATION

TOGETHER WE SHINE BRIGHT

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 the Cystinosis Research Foundation. Please note, this module:

- Takes approximately 45-60 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, 8:00am - 4:30pm (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.

☐ Cystinosis Research Foundation	☐ I do not give my permission

General Information

1. What is the participant's current weight? _____ lbs or _____ kg

2. What is the participant's current height? _____ feet _____ inches or _____ centimeters

3. What is the participant's natural hair color?

☐ Black	☐ Brown
☐ Blonde	☐ Red

Family History

4. If anyone in the participant's family died from cystinosis, what was the cause of death? If multiple family members died from cystinosis, select all that apply.

☐ Bleeding or blood clot	☐ Kidney failure

☐ Cancer	☐ Respiratory (lung) failure
☐ Cardiovascular (heart) disease	☐ Stroke
☐ Choking/aspiration	☐ Unsure
☐ Complications of diabetes	☐ Not applicable
☐ Infection	☐ Other
If "other", please specify:	
5. At approximately what age did the participant's relative with cystinosis die?	
Relative 1:	
Relative 2:	
Symptoms/Diagnosis	
6. What were the first cystinosis symptoms? (select all that apply):	
☐ Bone deformities	☐ Severe thirst
☐ Frequent urination	☐ Vomiting
☐ Loss of appetite	☐ Unusual urine odor
☐ Muscle weakness	☐ Weight loss
☐ Poor growth	☐ Unsure
☐ Problems walking	☐ None
☐ Sensitivity to light	☐ Other
If "other", please specify:	
7. How was the cystinosis diagnosis made?	
☐ Cystine measurement	☐ Slit lamp eye exam

☐ DNA test	☐ Unsure
☐ Prenatal cysteine measurement	

8. Has genetic (DNA) testing for cystinosis been performed?

☐ Yes	☐ No	☐ Unsure

Cysteamine Therapy

9. Was the participant ever treated with cysteamine (Cystagon®, Procysbi® or earlier versions of the medication)?
Cysteamine is the active ingredient in cystinosis drug therapies. Cystagon® and Procysbi® are brands of oral cysteamine therapy currently on the market. These questions refer to all oral forms of cysteamine only, and do not refer to cysteamine eye drops.

☐ Yes	☐ No	☐ Unsure

10. If the participant has not been treated with cysteamine (Cystagon®, Procysbi® or earlier versions), why not?

☐ Do not know where to get Cystagon®, Procysbi®

☐ Cannot afford Cystagon®, Procysbi®

☐ Cystagon®, Procysbi® are not available

Cysteamine Therapy Part II

These questions refer to all oral forms of cysteamine only, and do not refer to cysteamine eye drops.

11. Please select all forms of oral cysteamine the participant has used.

☐ Cystagon only

☐ Procysbi only

☐ Both Cystagon and Procysbi

☐ Other form, not Cystagon or Procysbi

12. At what age did the participant first receive oral cysteamine therapy (in any form)?
Years_____ months _____

13. How often over the past year did the participant, on average, miss a dose of cysteamine?

☐ Never	☐ Often (few times a week)

☐ Occasionally (once or twice a month)	☐ Very often (almost daily)
☐ Sometimes (once or twice a week)	☐ Unsure

14. Has the participant ever taken a "break" from taking cysteamine? This means the participant stopped completely (or nearly completely) taking cysteamine, not just missing a single dose periodically.

☐ Yes	☐ No	☐ Unsure

15. If the participant has taken cysteamine "breaks", what was the longest break?

☐ Days	☐ Year or more
☐ Weeks	☐ Unsure
☐ Months	☐ Not applicable

16. List any other factors that have caused the participant to miss a cysteamine dose or take differently than prescribed (select all that apply):

☐ Adherence to the frequent dosing schedule is too difficult	☐ The person has trouble swallowing pills
☐ Circumstances beyond the person's control (e.g. work or school demands) prevents the person from taking it	☐ There are too many capsules
☐ Forgets to take cysteamine	☐ Unpleasant odor
☐ Is too tired to take cysteamine	☐ Not applicable
☐ The person does not wake up from nightly sleep to take cysteamine (e.g. sleep past alarm)	☐ Other

If "other", please specify:

17. Has the participant experienced any of the following side effects from cysteamine? (select all that apply):

☐ Abdominal pain/discomfort/tenderness	☐ Gastrointestinal ulcers
☐ Bad odor	☐ Headaches
☐ Bad taste	☐ Heartburn
☐ Benign intracranial hypertension	☐ Low white blood cell count
☐ Bloating	☐ Nausea

☐ Bone and joint problems	☐ Skin rash
☐ Constipation	☐ Vomiting
☐ Decreased appetite	☐ None
☐ Diarrhea	☐ Unsure
☐ Elevated alkaline phosphatase	☐ Other
☐ Fatigue/sleepiness	
If "other", please specify:	

18. What form of cysteamine is the participant currently taking?

☐ An experimental version of cysteamine (i.e., a study drug was part of a clinical trial)	☐ Procysbi®
☐ Cystagon®	☐ Unsure

19. If the participant is currently not taking any form of cysteamine, why not?

☐ Cannot afford	☐ Cysteamine not available
☐ Cannot tolerate due to side effects or drug reaction	☐ Other
If "other", please specify:	

20. Has the participant been checking their cystine level while on treatment with cysteamine?

☐ Yes	☐ No	☐ Unsure

21. How many times over the last year has the participant had their cystine level checked?

☐ None over the last year	☐ Three times
☐ Once	☐ Four times
☐ Two times	☐ More than four times

22. What is the participant's most recent cystine level test result?

_____ cystine level (nmol/mg)	☐ Unsure

23. What test method was used to determine the participant's most recent cystine level?

☐ Mixed Leukocytes WBC ("old" method)	☐ Granulocyte ("new" method)	☐ Unsure

Cysteamine Therapy Part III (both Cystagon and Procysbi)

24. Which of the following best describes the participant's experience with oral cysteamine therapy?

☐ Used Cystagon® in the past but currently use Procysbi® and do not plan to use Cystagon® again

☐ Tried Procysbi® for a time but switched back to Cystagon® temporarily

☐ Tried Procysbi® for a time but switched back to Cystagon® and do not plan to use Procysbi® again

☐ None of the above

25. Which oral cysteamine therapy caused fewer side effects in the participant?

☐ Cystagon®	☐ Procysbi®	☐ No difference

Cystagon Therapy

Cystagon® is a brand of oral cysteamine therapy currently on the market, with recommended dosing every 6 hours. For the following questions, please answer about the participant's experience with Cystagon®

26. How many times per day does the participant usually take a dose of Cystagon®? _____

27. What is the participant's current dose of Cystagon®? (note, the small pills are 50mg each and the large pills are 150mg each) _____ mg	☐ Unsure

Procysbi Therapy

Procysbi® is a brand of oral cysteamine therapy currently on the market in the United States, with recommended dosing every 12 hours. For the following questions, please answer the participant's experience with Procysbi®.

28. How many times per day does the participant usually take a dose of Procysbi®? _____	
29. What is the participant's current dose of Procysbi®? (Note, the small pills are 25mg each and the large pills are 75mg each. Granulate packets are either 75mg (purple) or 300mg (blue)) _____ mg	☐ Unsure
Additional Medications	
30. Which of the following medications does the participant take? (select all that apply):	
☐ Vitamin D2 (ergocalciferol or Vitamin D3 (cholecalciferol)	☐ Anti-nausea ondansetron (Zofran), promethazine (Phenergan), prochlorperazine (Compazine), metoclopramide (Reglan)
☐ Calcitriol (Rocaltrol)	☐ Appetite stimulants, cyproheptadine (Periactin)
☐ Calcium (Calcium Carbonate or calcium acetate)	☐ Thyroid replacement (levothyroxine, Armour Thyroid)
☐ Potassium chloride (Klor-Con, K-Tab, K-Sol)	☐ Growth hormone
☐ Phosphorus (Phos-NaK, K-Phos Neutral, K-Phos) No.2, Phospha 250 Neutral, Virt-Phos 250 Neutral, Av-Phos 250 Neutral, Sodium Phosphate	☐ Testosterone (gel, patch, shots)
☐ Sodium bicarbonate	☐ Birth control pills
☐ Magnesium (Magnesium oxide, Uro-mag)	☐ Erythropoietin
☐ Potassium citrate (Cytra-K, Virtrate-K, Polycitra-K, Urocit-K)	☐ Megace (megestrol)
☐ Sodium citrate (Cytra-2, Virtrate-2, Oracit, Bicitra)	☐ Insulin
☐ Sodium and potassium citrate (Tricitrates, Polycitra and Cytra-3)	☐ Metformin
☐ Indomethacin (Indocin)	☐ Transplant medications (Prednisone, azathioprine (lmuran), cyclosporine, tacrolimus (Prograf or Astagra), mycophenolate (Cellcept or Myfortic), everolimus, sirolimus
☐ ACE inhibitor (Enalapril, Lisinopril, Ramipril, quinapril, benazepril, fosinopril)	☐ Cysteamine eye drops

☐ Angiotensin receptor blockers (ARB) (losartan, irbesartan, candesartan, valsartan)	☐ Muscle or Mitochondrial Support (CoQ 10 (Coenzyme Q), Vitamin B supplements, Carnitine, Creatine, L-Carnitine (Levocarnitine, Carnitor)
☐ Thiazide diuretic (Chlorothiazide (Diuril) or hydrochlorothiazide, chlorthalidone)	☐ Other blood pressure medications
☐ Proton pump inhibitors (omeprazole (Prilosec), esomeprazole (Nexium), lansoprazole (Prevacid))	☐ Other
☐ H2 Blocker (Zantac (ranitidine), Pepcid (famotidine))	

Kidney

Please answer the following questions about the participant's urine.

31. Approximately how many times per day does the participant usually urinate? _____

32. Does the participant have to wake up at night to urinate?

☐ Yes	☐ No

If "yes", how many times? _____

33. Does the participant have to wake up in the middle of the night to drink?

☐ Yes	☐ No

34. Does the participant experience nighttime bed wetting?

☐ Yes	☐ No

35. Does the participant have elevated amounts of protein in their urine (termed proteinuria or albuminuria)?

☐ Yes	☐ No	☐ Unsure

36. What was the participant's most recent serum or plasma creatinine level? Please enter in milligram/deciliter (United States) or Umol/L (other countries)

_____ milligram/deciliter	_____ Umol/L	☐ Unsure

37. What was the participant's most recent Glomerular filtration rate (GFR)?

For more information on calculating GFR for pediatrics:
https://www.kidney.org/professionals/KDOQI/gfr_calculatorPed

For more information on calculating GFR for adults:
https://www.kidney.org/professionals/kdoqi/gfr_calculator

_____ Units	☐ Unsure

38. What is the participant's most recent hemoglobin level? Please provide answer in gram/deciliter (United States) or fill in the g/L (other countries).

_____ gram/deciliter	_____ g/L	☐ Unsure
39. Has the participant ever had an Acute Kidney Injury (AKI), when kidney function decreased and then later improved?		
☐ Yes	☐ No	☐ Unsure
40. Has the participant ever required chronic dialysis?		
☐ Yes	☐ No	☐ Unsure
If "yes", at what age was dialysis started? _____ years		
41. If the participant has been dialyzed, what type(s) of dialysis has the participant used?		
☐ Hemodialysis	☐ Not applicable	
☐ Peritoneal		
42. In total, how long was the participant on dialysis? _____ years		

Kidney Transplant

43. Has the participant had a kidney transplant?	
☐ Yes	☐ No
44. If the participant has had a kidney transplant, how many transplants has the participant had?	
☐ One transplant	☐ Three transplants
☐ Two transplants	☐ More than three transplants
45. How old was the participant when they received the first transplant? _____ years	
46. How long did the first kidney transplant work? _____ years	
47. Please describe the first donated kidney:	
☐ From living blood relative	☐ From a deceased donor
☐ From living non-blood relative	☐ Unsure

48. How long was the participant on the National Kidney Transplant Waitlist for their first kidney transplant? _____ years _____ months

49. How old was the participant when they received the second transplant? _____ years

50. How long did the second kidney transplant work? _____ years

51. Please describe the second donated kidney:

☐ From living blood relative	☐ From a deceased donor
☐ From living non-blood relative	☐ Unsure

52. How long was the participant on the National Kidney Transplant Waitlist for their second kidney transplant? _____ years _____ months

53. How old was the participant when they received the third transplant? _____ years

54. How long did the third kidney transplant work? _____ years

55. Please describe the third donated kidney:

☐ From living blood relative	☐ From a deceased donor
☐ From living non-blood relative	☐ Unsure

56. How long was the participant on the National Kidney Transplant Waitlist for their third kidney transplant? _____ years _____ months

Musculoskeletal/Bone/Skin

57. Has the participant experienced any of the following bone-related problems? (select all that apply):

☐ Abnormal gait	☐ Other bone or cartilage problems
☐ Bone fracture	☐ Osteopenia and osteoporosis
☐ Bone pain	☐ Rickets
☐ Bow legged (genu varum)	☐ Short stature

☐ Chest wall deformity (i.e., breastbone is protuberant or depressed)	☐ Unsure
☐ Flat feet	☐ None of the above
☐ Knock knees (genu valgum)	

58. At what age did the physician, parent, or the participant first notice bone-related deformities?
_____ years

59. How many bone fractures has the participant had? *(if the participant has not had any bone fractures, please enter "0")* _____ fracture(s)

60. Has the participant ever had surgery to correct bone deformities?

☐ Yes	☐ Unsure
☐ No	☐ Not applicable

If "yes", please specify the type of surgery:

61. Has the participant ever noted unusual bruising (not related to an injury)?

☐ Yes	☐ No	☐ Unsure

62. Has the participant ever experienced any of the following other skin anomalies? (select all that apply):

☐ Cutaneous vascular lesions (abnormal blood vessel proliferation in skin)	☐ Other
☐ Papules (bumps)	☐ Not applicable
☐ Striae (stretch marks)	

If "other", please specify:

Please rate the following categories according to the participant's abilities. *If the participant is not old enough for some of the categories below, please select "Not applicable"*

63. Reaching above head

☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable

64. Carrying bags with groceries or other material

☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable

65. Difficulty opening jars or lids that other people do not have trouble with			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
66. Using a fork or knife			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
67. Tying shoelaces/putting on shoes			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
68. Brushing teeth			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
69. Clipping nails			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
70. Buttoning shirts			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
71. Tearing paper			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
72. Typing			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
73. Writing			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
74. Standing from a low seated position			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
75. Walking on uneven surfaces			

☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
76. Climbing stairs			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
77. Eating at a normal pace			
☐ No difficulty	☐ Some difficulty	☐ A lot of difficulty	☐ Not applicable
78. Does the participant have difficulty swallowing?			
☐ Yes	☐ No	☐ Unsure	
If "yes", how many years has the participant had difficulty swallowing? _____ years			
79. Has the participant experienced muscle weakness in any of the following areas? (select all that apply):			
☐ Abdomen		☐ Legs	
☐ Arms		☐ Neck	
☐ Chest		☐ Shoulders	
☐ Face/mouth/throat		☐ Unsure	
☐ Hand		☐ None	
80. If muscle weakness has been experienced, at what age did the participant start to experience muscle weakness? _____ years			
81. Has the muscle weakness impacted the participant's ability to function independently?			
☐ Yes	☐ No	☐ Not applicable	
82. Has the participant experienced difficulties with speech due to weakened vocal muscles?			
☐ Yes		☐ No	

83. Have speech difficulties significantly impacted the participant's ability to communicate with others?

☐ Yes	☐ No	☐ Not applicable

Gastrointestinal

84. Has the participant any of the following gastrointestinal problems over the last year? (select all that apply):

☐ Abdominal Pain	☐ Heartburn
☐ Constipation	☐ Liver disease
☐ Decreased appetite	☐ Nausea/Vomiting
☐ Diarrhea	☐ Pain associated with swallowing
☐ Difficulty swallowing (choking, gagging, coughing when swallowing)	☐ Peptic ulcers
☐ Does food get stuck while swallowing	☐ Other
☐ Gastroparesis	

85. If the participant has had a feeding tube, what type is/was it?

☐ Gastric-tube (G-tube)	☐ Nasoduodenal tube
☐ Gastrojejunal tube (G-J tube)	☐ Nasogastric tube
☐ Jejunal tube (J-tube)	☐ Not applicable

86. Has the participant ever been diagnosed with liver disease?

☐ Yes	☐ No	☐ Unsure

Endocrine/Reproductive

87. If the participant has been diagnosed with hypothyroidism (low thyroid function), at what age were they diagnosed?

_____ years _____ months	☐ Not applicable

88. If the participant has been diagnosed with diabetes mellitus, at what age was the participant diagnosed?		
_____ years	☐ Not applicable	
89. Has the participant experienced growth (height) delay?		
☐ Yes	☐ No	☐ Unsure
90. At what age did the participant's physician or parent notice poor growth?		
_____ years _____ months	☐ Not applicable	
91. If the participant has been treated with growth hormones, at what age was the treatment initiated?		
_____ years _____ months	☐ Not applicable	
92. For participant's who have stopped treatment with growth hormones, approximately how many years in total did the participant take growth hormones?		
_____ years	☐ Has not stopped (continues using growth hormone)	☐ Not applicable
93. At what age did the participant show signs of puberty? (For females, indicate the age that menstrual period began; for boys indicate the age that facial or body hair appeared).		
_____ years	☐ Not applicable	
94. If the participant is female, how many pregnancies has the participant had?		
_____ pregnancy(ies)	☐ Not applicable	
95. How many pregnancy(ies) resulted in a live birth?		
_____ pregnancy(ies)	☐ Not applicable	
96. If the participant is male, does the participant have a biological child?		
☐ Yes	☐ No	☐ Not applicable
97. Has the participant ever used "assisted reproductive technology" for example, in vitro fertilization, testicular or ovarian surgery to harvest sperm/egg?		
☐ Yes	☐ No	

Respiratory/Cardiovascular

98. Has the participant ever been diagnosed with decreased lung function?

☐ Yes	☐ No

99. Has the participant ever aspirated/inhaled food or liquid into their lungs?

☐ Yes	☐ No	☐ Unsure

100. Has the participant ever been diagnosed with pneumonia?

☐ Yes	☐ No	☐ Unsure

101. Has the participant ever been diagnosed with any of the following conditions? (select all that apply):

☐ Abnormal heart rhythm	☐ Hypertension
☐ Coronary artery disease	☐ Other cardiac disease
☐ Heart failure	☐ None of the above
☐ Heart murmur	

If "other", please specify:

102. Has the participant ever been diagnosed with sleep apnea?

☐ Yes	☐ No	☐ Unsure

Neurological

103. Please indicate if the participant has ever had problems with any of the following? (select all that apply):

☐ Autism spectrum disorder	☐ Pseudotumor cerebri (intracranial hypertension)
☐ Chiari malformation	☐ Seizures (convulsions)
☐ Dementia	☐ Severe headaches/migraines
☐ Developmental delay	☐ Stoke

☐ Hyperactivity or attention deficit (hyperactivity) disorder (ADHD, ADD)	☐ Transient Ischemic Attack (TIA)
☐ Learning difficulties	☐ Other
☐ Memory loss	☐ Unsure
☐ Problems with coordination	

104. If the participant has had migraine headaches, how often does the participant experience a migraine headache?

☐ Daily	☐ Less than once a month
☐ Weekly	☐ Not applicable
☐ Monthly	

105. Please select from the following if the participant requires or has required special instruction in the following subjects. (select all that apply):

☐ Math	☐ Writing
☐ Reading	☐ Other
☐ Spelling	

106. Please select from the following if the participant has had therapies or other interventions. (select all that apply):

☐ Behavioral therapy	☐ Physical therapy
☐ Individualized Education Plan (IEP)	☐ Speech/language therapy
☐ Medication	☐ Unsure
☐ Occupational therapy	☐ Not applicable

Ophthalmological/Visual

107. Please indicate if the participant has or has ever had any of the ophthalmologic symptoms listed below. (select all that apply):

☐ Blink or wink uncontrollably	☐ Light sensitivity or trouble seeing in the sunlight
☐ Blurry vision or trouble focusing	☐ Red eyes
☐ Crusting on your lashes	☐ Sandy or gritty or foreign body sensation
☐ Dry eye	☐ Teary eyes
☐ Eye pain or burning sensation	☐ Not applicable

108. Has the participant ever been diagnosed with any of the following eye conditions? (select all that apply):

☐ Cataract	☐ Retinopathy
☐ Dry eye	☐ Not applicable
☐ Glaucoma	

109. At what age did the participant's physician observe cystine crystals in the eyes? (select all that apply):

_____ years _____ months	☐ Unsure	☐ Not applicable

110. How would the participant rate their sensitivity to light (photophobia)?

☐ No sensitivity	☐ Severe
☐ Mild	☐ Very severe
☐ Moderate	

111. Has the participant ever used any form of cysteamine medication for their eyes?

☐ Yes	☐ No
112. Does the participant currently take cysteamine eye medication?	
☐ Yes	☐ No
113. If the participant takes cysteamine eye medication, select all forms the participant has ever used:	
☐ Cystaran® eye drops	☐ Other
☐ Cystadrops® eye gel	☐ None
☐ Compounded eye drops made by local pharmacist	
114. If the participant takes Cystaran® or compounded eye drops, how many doses on average does the participant take in each eye per day?	
_____	☐ Not applicable
115. If the participant has used Cystaran® or compounded eye drops, what side effects has the participant experienced? (select all that apply):	
☐ Blurry vision or vision loss	☐ Puffy eyes
☐ Crusting eyes	☐ Redness
☐ Eye pain/irritation	☐ Other
If "other", please specify:	
116. If the participant takes Cystadrops®, on average, how many doses do they take in each eye per day?	
_____	☐ Not applicable
117. If the participant takes Cystadrops®, what side effects has the participant experienced? (select all that apply):	
☐ Blurry vision or vision loss	☐ Puffy eyes

☐ Crusting eyes	☐ Redness
☐ Eye pain/irritation	☐ Other
If "other", please specify:	
118. How frequently is the participant seen by an eye care specialist?	
☐ Every 6 months	☐ As needed only
☐ Annually	☐ Does not see an eye care specialist
☐ Less than once a year	
119. Has the participant ever required corneal surgery or corneal transplant?	
☐ Yes	☐ No

Psychological/Quality of Life

120. Has the participant ever experienced any of the following? (select all that apply):		
☐ Anxiety	☐ Stress	
☐ Depression	☐ Other symptoms affecting mood or perception	
☐ Fatigue	☐ Not applicable	
121. Has the participant ever sought counseling or therapy?		
☐ Yes	☐ No	☐ Unsure
122. Has the participant ever been diagnosed with any of the following psychiatric disorders? (select all that apply):		
☐ Bipolar disorder	☐ Other	

☐ Generalized anxiety disorder	☐ None
☐ Major depressive disorder	
If "other", please specify:	
123. How much does pain and/or discomfort interfere with the participant's daily functioning?	
☐ Never	☐ Often
☐ Rarely	☐ Always
☐ Sometimes	☐ Unsure
124. Has cystinosis impacted the participant's school/work attendance?	
☐ Never	☐ Often
☐ Rarely	☐ Always
☐ Sometimes	☐ Unsure
125. Does the participant experience learning/memory/attention problems in school, at work, or in daily life?	
☐ Never	☐ Often
☐ Rarely	☐ Always
☐ Sometimes	☐ Unsure
126. What is the highest level of education the participant has completed?	
☐ No schooling completed	☐ Bachelor's degree
☐ Some high school or diploma	☐ Master's degree

☐ High school graduate	☐ Doctorate degree
☐ Professional/Vocational/Technical Training	
127. Is the participant currently employed?	
☐ Yes, full-time	☐ No
☐ Yes, part-time	☐ Not of working age
128. How much has cystinosis impacted the participant's decision or ability to have a significant other or partner?	
☐ Not at all	☐ Quite a bit
☐ A little bit	☐ Very much
☐ Somewhat	☐ Unsure
129. Does the participant experience any problems with sleep; going to sleep, staying asleep or sleeping too much?	
☐ Never	☐ Often
☐ Rarely	☐ Always
☐ Sometimes	☐ Unsure
130. How much has cystinosis impacted the participant's social interactions?	
☐ Not at all	☐ Quite a bit
☐ A little bit	☐ Very much
☐ Somewhat	☐ Unsure
131. How much has cystinosis impacted the participant's family interactions?	

☐ Not at all	☐ Quite a bit
☐ A little bit	☐ Very much
☐ Somewhat	☐ Unsure
132. To what degree has cystinosis created financial problems for the participant and/or their family?	
☐ Not at all	☐ Quite a bit
☐ A little bit	☐ Very much
☐ Somewhat	☐ Unsure
133. People with chronic disease may worry about many things. Please score how much the participant or the caregiver worries about each of the following. Use a scale of 1 for no worries to 5 for extreme and constant worry.	
Ability to have a home _____	Being a burden to your loved ones _____
Ability to live on your own _____	Developing the later stage complications of cystinosis _____
Ability to work and support yourself _____	Health insurance _____
Ability to have relationships like a significant other_____	Not living as long as you want to _____
Ability to have a child _____	The risk of passing on your condition to a child _____
Ability to raise a child _____	☐ Not applicable

134. How would the participant describe their daily quality of life?

- ☐ Excellent
- ☐ Mostly satisfied
- ☐ Mixed
- ☐ Mostly dissatisfied
- ☐ Terrible

Other Medical Issues

135. Has the participant ever experienced any of the following health issues? (select all that apply):

☐ Cancer	☐ Vasculitis
☐ Hearing impairment or loss	☐ Other
☐ High cholesterol	☐ None
☐ Non-malignant tumor	

If "other", please specify: