

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

<input type="checkbox"/> Cancer	<input type="checkbox"/> Respiratory (lung) failure
<input type="checkbox"/> Cardiovascular (heart) disease	<input type="checkbox"/> Stroke
<input type="checkbox"/> Choking/aspiration	<input type="checkbox"/> Unsure
<input type="checkbox"/> Complications of diabetes	<input type="checkbox"/> Not applicable
<input type="checkbox"/> Infection	<input type="checkbox"/> 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):

<input type="checkbox"/> Bone deformities	<input type="checkbox"/> Severe thirst
<input type="checkbox"/> Frequent urination	<input type="checkbox"/> Vomiting
<input type="checkbox"/> Loss of appetite	<input type="checkbox"/> Unusual urine odor
<input type="checkbox"/> Muscle weakness	<input type="checkbox"/> Weight loss
<input type="checkbox"/> Poor growth	<input type="checkbox"/> Unsure
<input type="checkbox"/> Problems walking	<input type="checkbox"/> None
<input type="checkbox"/> Sensitivity to light	<input type="checkbox"/> Other

If “other”, please specify:

7. How was the cystinosis diagnosis made?

<input type="checkbox"/> Cystine measurement	<input type="checkbox"/> Slit lamp eye exam
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<input type="checkbox"/> DNA test	<input type="checkbox"/> Unsure
<input type="checkbox"/> Prenatal cysteine measurement	

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

<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Unsure
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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.

<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Unsure
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10. If the participant has not been treated with cysteamine (Cystagon®, Procysbi® or earlier versions), why not?

<input type="checkbox"/> Do not know where to get Cystagon®, Procysbi®
<input type="checkbox"/> Cannot afford Cystagon®, Procysbi®
<input type="checkbox"/> 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.

<input type="checkbox"/> Cystagon only
<input type="checkbox"/> Procysbi only
<input type="checkbox"/> Both Cystagon and Procysbi
<input type="checkbox"/> 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?

<input type="checkbox"/> Never	<input type="checkbox"/> Often (few times a week)
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<input type="checkbox"/> Occasionally (once or twice a month)	<input type="checkbox"/> Very often (almost daily)
<input type="checkbox"/> Sometimes (once or twice a week)	<input type="checkbox"/> 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.	
<input type="checkbox"/> Yes	<input type="checkbox"/> No
<input type="checkbox"/> Unsure	
15. If the participant has taken cysteamine “breaks”, what was the longest break?	
<input type="checkbox"/> Days	<input type="checkbox"/> Year or more
<input type="checkbox"/> Weeks	<input type="checkbox"/> Unsure
<input type="checkbox"/> Months	<input type="checkbox"/> 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):	
<input type="checkbox"/> Adherence to the frequent dosing schedule is too difficult	<input type="checkbox"/> The person has trouble swallowing pills
<input type="checkbox"/> Circumstances beyond the person’s control (e.g. work or school demands) prevents the person from taking it	<input type="checkbox"/> There are too many capsules
<input type="checkbox"/> Forgets to take cysteamine	<input type="checkbox"/> Unpleasant odor
<input type="checkbox"/> Is too tired to take cysteamine	<input type="checkbox"/> Not applicable
<input type="checkbox"/> The person does not wake up from nightly sleep to take cysteamine (e.g. sleep past alarm)	<input type="checkbox"/> Other
If “other”, please specify:	
17. Has the participant experienced any of the following side effects from cysteamine? (select all that apply):	
<input type="checkbox"/> Abdominal pain/discomfort/tenderness	<input type="checkbox"/> Gastrointestinal ulcers
<input type="checkbox"/> Bad odor	<input type="checkbox"/> Headaches
<input type="checkbox"/> Bad taste	<input type="checkbox"/> Heartburn
<input type="checkbox"/> Benign intracranial hypertension	<input type="checkbox"/> Low white blood cell count
<input type="checkbox"/> Bloating	<input type="checkbox"/> Nausea

<input type="checkbox"/> Bone and joint problems	<input type="checkbox"/> Skin rash	
<input type="checkbox"/> Constipation	<input type="checkbox"/> Vomiting	
<input type="checkbox"/> Decreased appetite	<input type="checkbox"/> None	
<input type="checkbox"/> Diarrhea	<input type="checkbox"/> Unsure	
<input type="checkbox"/> Elevated alkaline phosphatase	<input type="checkbox"/> Other	
<input type="checkbox"/> Fatigue/sleepiness		
If “other”, please specify:		
18. What form of cysteamine is the participant currently taking?		
<input type="checkbox"/> An experimental version of cysteamine (i.e., a study drug was part of a clinical trial)	<input type="checkbox"/> Procysbi®	
<input type="checkbox"/> Cystagon®	<input type="checkbox"/> Unsure	
19. If the participant is currently not taking any form of cysteamine, why not?		
<input type="checkbox"/> Cannot afford	<input type="checkbox"/> Other	
<input type="checkbox"/> Cannot tolerate due to side effects or drug reaction	<input type="checkbox"/> Not applicable	
<input type="checkbox"/> Cysteamine not available		
If “other”, please specify:		
20. Has the participant been checking their cystine level while on treatment with cysteamine?		
<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Unsure
21. How many times over the last year has the participant had their cystine level checked?		
<input type="checkbox"/> None over the last year	<input type="checkbox"/> Three times	
<input type="checkbox"/> Once	<input type="checkbox"/> Four times	
<input type="checkbox"/> Two times	<input type="checkbox"/> 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®

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 Procsybi®? _____

29. What is the participant's current dose of Procsybi®? (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 (Imuran), cyclosporine, tacrolimus (Prograf or Astagra), mycophenolate (Cellcept or Myfortic), everolimus, sirolimus)

ACE inhibitor (Enalapril, Lisinopril, Ramipril, quinapril, benazepril, fosinopril)

Cysteamine eye drops

<input type="checkbox"/> Angiotensin receptor blockers (ARB) (losartan, irbesartan, candesartan, valsartan)	<input type="checkbox"/> Muscle or Mitochondrial Support (CoQ 10 (Coenzyme Q), Vitamin B supplements, Carnitine, Creatine, L-Carnitine (Levocarnitine, Carnitor)
<input type="checkbox"/> Thiazide diuretic (Chlorothiazide (Diuril) or hydrochlorothiazide, chlorthalidone)	<input type="checkbox"/> Other blood pressure medications
<input type="checkbox"/> Proton pump inhibitors (omeprazole (Prilosec), esomeprazole (Nexium), lansoprazole (Prevacid))	<input type="checkbox"/> Other
<input type="checkbox"/> 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

_____ mL/min/1.73 m²

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	<input type="checkbox"/> Unsure
39. Has the participant ever had an Acute Kidney Injury (AKI), when kidney function decreased and then later improved?		
<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Unsure
40. Has the participant ever required chronic dialysis?		
<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> 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?		
<input type="checkbox"/> Hemodialysis	<input type="checkbox"/> Not applicable	
<input type="checkbox"/> Peritoneal		
42. In total, how long was the participant on dialysis? _____years		
Kidney Transplant		
43. Has the participant had a kidney transplant?		
<input type="checkbox"/> Yes	<input type="checkbox"/> No	
44. If the participant has had a kidney transplant, how many transplants has the participant had?		
<input type="checkbox"/> One transplant	<input type="checkbox"/> Three transplants	
<input type="checkbox"/> Two transplants	<input type="checkbox"/> 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:		
<input type="checkbox"/> From living blood relative	<input type="checkbox"/> From a deceased donor	
<input type="checkbox"/> From living non-blood relative	<input type="checkbox"/> 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

<input type="checkbox"/> Chest wall deformity (i.e., breastbone is protuberant or depressed)	<input type="checkbox"/> Unsure		
<input type="checkbox"/> Flat feet	<input type="checkbox"/> None of the above		
<input type="checkbox"/> 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?			
<input type="checkbox"/> Yes	<input type="checkbox"/> Unsure		
<input type="checkbox"/> No	<input type="checkbox"/> Not applicable		
If "yes", please specify the type of surgery:			
61. Has the participant ever noted unusual bruising (not related to an injury)?			
<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Unsure	
62. Has the participant ever experienced any of the following other skin anomalies? (select all that apply):			
<input type="checkbox"/> Cutaneous vascular lesions (abnormal blood vessel proliferation in skin)	<input type="checkbox"/> Other		
<input type="checkbox"/> Papules (bumps)	<input type="checkbox"/> Not applicable		
<input type="checkbox"/> 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			
<input type="checkbox"/> No difficulty	<input type="checkbox"/> Some difficulty	<input type="checkbox"/> A lot of difficulty	<input type="checkbox"/> Not applicable
64. Carrying bags with groceries or other material			
<input type="checkbox"/> No difficulty	<input type="checkbox"/> Some difficulty	<input type="checkbox"/> A lot of difficulty	<input type="checkbox"/> Not applicable

65. Difficulty opening jars or lids that other people do not have trouble with

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> No difficulty | <input type="checkbox"/> Some difficulty | <input type="checkbox"/> A lot of difficulty | <input type="checkbox"/> Not applicable |
|--|--|--|---|

66. Using a fork or knife

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> No difficulty | <input type="checkbox"/> Some difficulty | <input type="checkbox"/> A lot of difficulty | <input type="checkbox"/> Not applicable |
|--|--|--|---|

67. Tying shoelaces/putting on shoes

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> No difficulty | <input type="checkbox"/> Some difficulty | <input type="checkbox"/> A lot of difficulty | <input type="checkbox"/> Not applicable |
|--|--|--|---|

68. Brushing teeth

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> No difficulty | <input type="checkbox"/> Some difficulty | <input type="checkbox"/> A lot of difficulty | <input type="checkbox"/> Not applicable |
|--|--|--|---|

69. Clipping nails

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> No difficulty | <input type="checkbox"/> Some difficulty | <input type="checkbox"/> A lot of difficulty | <input type="checkbox"/> Not applicable |
|--|--|--|---|

70. Buttoning shirts

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> No difficulty | <input type="checkbox"/> Some difficulty | <input type="checkbox"/> A lot of difficulty | <input type="checkbox"/> Not applicable |
|--|--|--|---|

71. Tearing paper

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> No difficulty | <input type="checkbox"/> Some difficulty | <input type="checkbox"/> A lot of difficulty | <input type="checkbox"/> Not applicable |
|--|--|--|---|

72. Typing

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> No difficulty | <input type="checkbox"/> Some difficulty | <input type="checkbox"/> A lot of difficulty | <input type="checkbox"/> Not applicable |
|--|--|--|---|

73. Writing

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> No difficulty | <input type="checkbox"/> Some difficulty | <input type="checkbox"/> A lot of difficulty | <input type="checkbox"/> Not applicable |
|--|--|--|---|

74. Standing from a low seated position

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> No difficulty | <input type="checkbox"/> Some difficulty | <input type="checkbox"/> A lot of difficulty | <input type="checkbox"/> Not applicable |
|--|--|--|---|

75. Walking on uneven surfaces

<input type="checkbox"/> No difficulty	<input type="checkbox"/> Some difficulty	<input type="checkbox"/> A lot of difficulty	<input type="checkbox"/> Not applicable
76. Climbing stairs			
<input type="checkbox"/> No difficulty	<input type="checkbox"/> Some difficulty	<input type="checkbox"/> A lot of difficulty	<input type="checkbox"/> Not applicable
77. Eating at a normal pace			
<input type="checkbox"/> No difficulty	<input type="checkbox"/> Some difficulty	<input type="checkbox"/> A lot of difficulty	<input type="checkbox"/> Not applicable
78. Does the participant have difficulty swallowing?			
<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> 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):			
<input type="checkbox"/> Abdomen	<input type="checkbox"/> Legs		
<input type="checkbox"/> Arms	<input type="checkbox"/> Neck		
<input type="checkbox"/> Chest	<input type="checkbox"/> Shoulders		
<input type="checkbox"/> Face/mouth/throat	<input type="checkbox"/> Unsure		
<input type="checkbox"/> Hand	<input type="checkbox"/> 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?			
<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Not applicable	
82. Has the participant experienced difficulties with speech due to weakened vocal muscles?			
<input type="checkbox"/> Yes	<input type="checkbox"/> No		

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

Yes

No

Not applicable

Gastrointestinal

**84. Has the participant experienced 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 (feeling full quickly)

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	<input type="checkbox"/> Not applicable	
89. Has the participant experienced growth (height) delay?		
<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Unsure
90. At what age did the participant's physician or parent notice poor growth?		
_____years_____months	<input type="checkbox"/> Not applicable	
91. If the participant has been treated with growth hormones, at what age was the treatment initiated?		
_____years _____months	<input type="checkbox"/> 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	<input type="checkbox"/> Has not stopped (continues using growth hormone)	<input type="checkbox"/> 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	<input type="checkbox"/> Not applicable	
94. If the participant is female, how many pregnancies has the participant had?		
_____pregnancy(ies)	<input type="checkbox"/> Not applicable	
95. How many pregnancy(ies) resulted in a live birth?		
_____pregnancy(ies)	<input type="checkbox"/> Not applicable	
96. If the participant is male, does the participant have a biological child?		
<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Not applicable
97. Has the participant ever used "assisted reproductive technology" for example, in vitro fertilization, testicular or ovarian surgery to harvest sperm/egg?		
<input type="checkbox"/> Yes	<input type="checkbox"/> 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

Stroke

<input type="checkbox"/> Hyperactivity or attention deficit (hyperactivity) disorder (ADHD, ADD)	<input type="checkbox"/> Transient Ischemic Attack (TIA)
<input type="checkbox"/> Learning difficulties	<input type="checkbox"/> Other
<input type="checkbox"/> Memory loss	<input type="checkbox"/> Unsure
<input type="checkbox"/> Problems with coordination	

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

<input type="checkbox"/> Daily	<input type="checkbox"/> Less than once a month
<input type="checkbox"/> Weekly	<input type="checkbox"/> Not applicable
<input type="checkbox"/> Monthly	

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

<input type="checkbox"/> Math	<input type="checkbox"/> Writing
<input type="checkbox"/> Reading	<input type="checkbox"/> Other
<input type="checkbox"/> Spelling	

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

<input type="checkbox"/> Behavioral therapy	<input type="checkbox"/> Physical therapy
<input type="checkbox"/> Individualized Education Plan (IEP)	<input type="checkbox"/> Speech/language therapy
<input type="checkbox"/> Medication	<input type="checkbox"/> Unsure
<input type="checkbox"/> Occupational therapy	<input type="checkbox"/> 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):

<input type="checkbox"/> Blink or wink uncontrollably	<input type="checkbox"/> Light sensitivity or trouble seeing in the sunlight
<input type="checkbox"/> Blurry vision or trouble focusing	<input type="checkbox"/> Red eyes
<input type="checkbox"/> Crusting on your lashes	<input type="checkbox"/> Sandy or gritty or foreign body sensation
<input type="checkbox"/> Dry eye	<input type="checkbox"/> Teary eyes
<input type="checkbox"/> Eye pain or burning sensation	<input type="checkbox"/> Not applicable

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

<input type="checkbox"/> Cataract	<input type="checkbox"/> Retinopathy
<input type="checkbox"/> Dry eye	<input type="checkbox"/> Not applicable
<input type="checkbox"/> Glaucoma	

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

_____ years _____ months	<input type="checkbox"/> Unsure	<input type="checkbox"/> Not applicable
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110. How would the participant rate their sensitivity to light (photophobia)?

<input type="checkbox"/> No sensitivity	<input type="checkbox"/> Severe
<input type="checkbox"/> Mild	<input type="checkbox"/> Very severe
<input type="checkbox"/> Moderate	

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

<input type="checkbox"/> Yes	<input type="checkbox"/> No
112. Does the participant currently take cysteamine eye medication?	
<input type="checkbox"/> Yes	<input type="checkbox"/> No
113. If the participant takes cysteamine eye medication, select all forms the participant has ever used:	
<input type="checkbox"/> Cystaran® eye drops	<input type="checkbox"/> Other
<input type="checkbox"/> Cystadrops® eye gel	<input type="checkbox"/> None
<input type="checkbox"/> 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?	
_____	<input type="checkbox"/> Not applicable
115. If the participant has used Cystaran® or compounded eye drops, what side effects has the participant experienced? (select all that apply):	
<input type="checkbox"/> Blurry vision or vision loss	<input type="checkbox"/> Redness
<input type="checkbox"/> Crusting eyes	<input type="checkbox"/> Other
<input type="checkbox"/> Eye pain/irritation	<input type="checkbox"/> Not applicable
<input type="checkbox"/> Puffy eyes	
If "other", please specify:	

116. If the participant takes Cystadrops®, on average, how many doses do they take in each eye per day?	
_____	<input type="checkbox"/> Not applicable
117. If the participant takes Cystadrops®, what side effects has the participant experienced? (select all that apply):	

<input type="checkbox"/> Blurry vision or vision loss	<input type="checkbox"/> Redness
<input type="checkbox"/> Crusting eyes	<input type="checkbox"/> Other
<input type="checkbox"/> Eye pain/irritation	<input type="checkbox"/> Not applicable
<input type="checkbox"/> Puffy eyes	

If "other", please specify:

118. How frequently is the participant seen by an eye care specialist?

<input type="checkbox"/> Every 6 months	<input type="checkbox"/> As needed only
<input type="checkbox"/> Annually	<input type="checkbox"/> Does not see an eye care specialist
<input type="checkbox"/> Less than once a year	

119. Has the participant ever required corneal surgery or corneal transplant?

<input type="checkbox"/> Yes	<input type="checkbox"/> No
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Psychological/Quality of Life

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

<input type="checkbox"/> Anxiety	<input type="checkbox"/> Stress
<input type="checkbox"/> Depression	<input type="checkbox"/> Other symptoms affecting mood or perception
<input type="checkbox"/> Fatigue	<input type="checkbox"/> Not applicable

121. Has the participant ever sought counseling or therapy?

<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Unsure
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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?

<input type="checkbox"/> No schooling completed	<input type="checkbox"/> Bachelor's degree
<input type="checkbox"/> Currently enrolled in elementary/secondary school	<input type="checkbox"/> Master's degree
<input type="checkbox"/> Some high school or diploma	<input type="checkbox"/> Doctorate degree
<input type="checkbox"/> High school graduate	
<input type="checkbox"/> Professional/Vocational/Technical Training	
127. Is the participant currently employed?	
<input type="checkbox"/> Yes, full-time	<input type="checkbox"/> No
<input type="checkbox"/> Yes, part-time	<input type="checkbox"/> Not of working age
128. How much has cystinosis impacted the participant's decision or ability to have a significant other or partner?	
<input type="checkbox"/> Not at all	<input type="checkbox"/> Quite a bit
<input type="checkbox"/> A little bit	<input type="checkbox"/> Very much
<input type="checkbox"/> Somewhat	<input type="checkbox"/> Unsure
129. Does the participant experience any problems with sleep; going to sleep, staying asleep or sleeping too much?	
<input type="checkbox"/> Never	<input type="checkbox"/> Often
<input type="checkbox"/> Rarely	<input type="checkbox"/> Always
<input type="checkbox"/> Sometimes	<input type="checkbox"/> Unsure
130. How much has cystinosis impacted the participant's social interactions?	
<input type="checkbox"/> Not at all	<input type="checkbox"/> Quite a bit

<input type="checkbox"/> A little bit	<input type="checkbox"/> Very much
<input type="checkbox"/> Somewhat	<input type="checkbox"/> Unsure
131. How much has cystinosis impacted the participant's family interactions?	

<input type="checkbox"/> Not at all	<input type="checkbox"/> Quite a bit
<input type="checkbox"/> A little bit	<input type="checkbox"/> Very much
<input type="checkbox"/> Somewhat	<input type="checkbox"/> Unsure

132. To what degree has cystinosis created financial problems for the participant and/or their family?

<input type="checkbox"/> Not at all	<input type="checkbox"/> Quite a bit
<input type="checkbox"/> A little bit	<input type="checkbox"/> Very much
<input type="checkbox"/> Somewhat	<input type="checkbox"/> 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 _____	<input type="checkbox"/> 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: