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 PFIC Network, Inc. Please note, this module:

- Takes approximately 30 minutes to complete
- Will refer to the person with the diagnosis as "**the participant**"
- You may see some questions that look like this "PFIC [yes/no] or Transplant [yes/no]. In these questions, please circle yes next to whether the patient is currently experiencing PFIC or has been transplanted related to PFIC.
- Can be updated at any time by logging in to the CoRDS online portal or by contacting CoRDS personnel
- Throughout the questionnaire, please describe the age in months for patients under 36 months, and describe the age in years for patients older than 36 months.

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.

☐ PFIC Network	☐ I do not give my permission to share my information with PFIC Network

Demographics

1. If you are the participant's parent or legally authorized representative, please indicate your relationship with the participant.

☐ Mother	☐ Not applicable
☐ Father	☐ Other
☐ Legal guardian	

If "other", please specify:

2. What is the participant's weight? (use whichever value is known to you)	
______________lbs	____________________kgs
3. What is the participant's height? (use whichever value is known to you)	
________________in	___________________cm
4. If the participant is age 5 or older, does the participant currently attend school?	
☐ Yes	☐ No
☐ Homeschooled	☐ Not Applicable
5. If "yes", does the participant require any additional assistance to facilitate their educational needs (ie. A learning plan, supplemental instruction, an educational assistant)	
☐ Yes	☐ No
If "yes", please specify:	

Diagnosis		
6. Did the participant undergo genetic testing as part of the evaluation for PFIC?		
☐ Yes	☐ No	☐ I don't know
7. If "yes", did the genetic testing lead to a diagnosis? (please see instructions at the end of this survey for instructions on how to upload genetic testing results if available)		
☐ Yes	☐ No	☐ I don't know
If "yes", please specify:		

8. What subtype of PFIC does the participant have?	
☐ Type 1 (FIC 1)	☐ Type 2 (BSEP)
☐ Type 3 (MDR 3)	☐ Type 4 (TJP2)

☐ Type 5 (MYO5B)	☐ FXR
☐ BRIC	☐ Unknown
☐ Other	
If "other", please specify which gene is detected:	
If the participant has a subtype of "BRIC", please specify:	
☐ BRIC 1 (FIC1)	☐ Unknown
☐ BRIC 2 (BSEP)	

Symptoms

**Note, if patient has a diagnosis of BRIC, please fill in the remainder of the survey with your experiences during an episode of symptoms.*

9. **Has the participant ever experienced any of the following problems? (select all that apply)**

☐ Acholic stools (white or grey poop)	☐ Gallstones
☐ Ascites	☐ Gastrointestinal bleeding
☐ Bleeding	☐ Hepatocellular carcinoma (liver cancer)
☐ Bruising	☐ Intellectual disability
☐ Cholestatis	☐ Jaundice
☐ Cirrhosis	☐ Pancreatitis
☐ Deafness	☐ Pruritus (itching/scratching)
☐ Developmental delay	☐ Rickets

☐ Diarrhea	☐ Sleep problems
☐ Difficulty with feeding or eating	☐ Varices
☐ Enlarged spleen	☐ Vitamin deficiencies
☐ Failure to thrive	☐ Other
☐ Fractures or breaks	
If “other”, please specify:	

10. Did the participant have neonatal cholestasis?		
☐ Yes	☐ No	☐ I don’t know

11. Does the participant currently have or use any of the following nutritional interventions? (select all that apply)	
☐ G-Tube (surgically placed tube in abdomen)	☐ Special formula (please specify in comments)
☐ MCT oil	☐ Other
☐ NG tube (temporary tube in nose)	
If applicable, please provide additional information on special formulas and MCT oil:	
If “other”, please specify:	
12. On average, how often does the participant wake up through the night?	
☐ Every hour	☐ Every 1-3 hours

☐ Every 3-5 hours	☐ Participant sleeps all night

The next four questions are related to pruritus (itching). If the participant has never experienced pruritus (itching) please move on to question 17.

13. Please rate the severity of the participant's itch at its worst point in the participant's lifetime (1=little itch, 10=extreme)

☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6	☐ 7	☐ 8	☐ 9	☐ 10

14. Please rate the severity of the participant's itch right now. (0=no itch, 10=extreme)

☐ 0	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6	☐ 7	☐ 8	☐ 9	☐ 10

15. Please rate the impact that the participant's itch has on the quality of life of the participant and other's in the participant's family?
1=no impact, itching does not interfere with quality of life.
5=severe impact, quality of life is severely affected due to participant's itching.

Participant	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ Not Applicable
Mother	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ Not Applicable
Father	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ Not Applicable
Other Caregiver	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ Not Applicable
Siblings	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ Not Applicable
Friends	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ Not Applicable
Other Relatives	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ Not Applicable

16. How does the participant's itch interfere with the participant's quality of life? (Select all that apply):

☐ Difficulty eating due to itch	☐ Itching to the point of bleeding (self-mutilation)

☐ Difficulty grooming due to itch (dressing, baths, etc.)	☐ Lack of sleep or poor sleep due to itch
☐ Difficulty playing with friends	☐ Low self esteem
☐ Fatigue due to itch	☐ Others in our life don't understand the impact of the itch
☐ Feelings of hopelessness due to itch	☐ Pain due to itch
☐ Inability to concentrate at school	☐ The patient's healthcare providers don't understand the impact of the itch
☐ Inconsolable crying due to itch	☐ Other
If "other", please specify:	

Treatment

17. What medication is the participant taking or has previously taken for PFIC?
If the participant has never taken medication, please move on to question 18.

A clinical trial drug	☐ Taking now	☐ Taken in the past	☐ Not applicable
Rifampicin	☐ taking now	☐ Taken in the past	☐ Not applicable
Ursodiol (UDCA)	☐ Taking now	☐ Taken in the past	☐ Not applicable
Vitamin A	☐ Taking now	☐ Taken in the past	☐ Not applicable
Vitamin D	☐ Taking now	☐ Taken in the past	☐ Not applicable
Vitamin E	☐ Taking now	☐ Taken in the past	☐ Not applicable
Vitamin K	☐ Taking now	☐ Taken in the past	☐ Not applicable
Multivitamin	☐ Taking now	☐ Taken in the past	☐ Not applicable
Naltrexone	☐ Taking now	☐ Taken in the past	☐ Not applicable

Cholestyramine	☐ Taking now	☐ Taken in the past	☐ Not applicable
Phenobarbital	☐ Taking now	☐ Taken in the past	☐ Not applicable
Antihistamine (Benadryl, hydroxyzine, etc.)	☐ Taking now	☐ Taken in the past	☐ Not applicable
Odansetron	☐ Taking now	☐ Taken in the past	☐ Not applicable
Steroid (hydrocortisone, prednisone, etc.)	☐ Taking now	☐ Taken in the past	☐ Not applicable
Sertroline	☐ Taking now	☐ Taken in the past	☐ Not applicable
Carbamazepine	☐ Taking now	☐ Taken in the past	☐ Not applicable
Tacrolimus	☐ Taking now	☐ Taken in the past	☐ Not applicable
Sirolimus	☐ Taking now	☐ Taken in the past	☐ Not applicable
Mycophenalate	☐ Taking now	☐ Taken in the past	☐ Not applicable
Other:________________	☐ Taking now	☐ Taken in the past	☐ Not applicable
Other:_________________	☐ Taking now	☐ Taken in the past	☐ Not applicable

18. Has the participant ever undergone any of the following surgical procedures? (select all that apply):

☐ Cholecysto-appendicostomy	☐ Partial **External** biliary diversion
☐ Kasai Procedure (hepatoportoenterostomy)	☐ Partial **Internal** biliary diversion
☐ Ileal Exclusion	☐ Unknown
☐ Liver Transplant	☐ Other

☐ No surgery	
If "other", please specify:	
19. Please specify the participant's age at each surgical intervention (please describe age in months under 36 months; describe age in years for participants older than 36 months).	
Surgical Intervention	**Age**
___________________________	Years_____________ Months _____________
___________________________	Years_____________ Months _____________
___________________________	Years_____________ Months _____________
___________________________	Years_____________ Months _____________
20. If the participant received a liver transplant, what is your understanding of the primary reason(s)? (select all that apply): *(if the patient has not received a liver transplant, please move on to question 22)*	
☐ End stage liver disease due to PFIC	☐ Portal hypertension
☐ Growth Failure	☐ Unknown
☐ Intractable pruritus (uncontrolled itching)	☐ Other
☐ Liver Cancer	
If "other", please specify:	
21. If the participant received a liver transplant, what type of graft (organ) was used? (Select all that apply):	
☐ Deceased Donor	☐ Living related donor
☐ Living unrelated donor, known	☐ Unknown

☐ Living unrelated donor, anonymous	☐ Other
If "other", please specify:	
22. In the past year, how often has the participant been seen by a doctor for his/her condition?	
☐ Never	☐ Every 3 months
☐ Twice per year (every 6 months)	☐ Other
☐ Once per year	
If "other", please specify:	
23. Has the participant ever seen a specialist for his/her PFIC (a doctor who specializes in liver disease)?	
☐ Yes	☐ No
If the participant sees a specialist for his/her PFIC, please provide the following information:	
Name of Hospital/Clinic:	
City:	
State:	
Country if not in the United States:	
24. If the participant sees any other specialists, please select from the following:	
☐ Counselor	☐ Psychologist
☐ Developmental Specialist	☐ Speech Therapist
☐ Dietician	☐ Social Worker

☐ Occupational Therapist	☐ Other
☐ Physical Therapist	
If “other”, please specify:	
25. How often over the past 12 months, has the participant been ADMITTED to the hospital for at least one night to treat complications related to their condition?	
☐ The patient has not been admitted to the hospital in the past 12 months	☐ Three times
☐ One time	☐ Four times
☐ Two times	☐ Greater than 4 times
26. Was the participant covered by any form of insurance in the last 12 months?	
☐ No health insurance	☐ Yes, government health insurance
☐ Yes, private insurance	☐ Yes, government and private insurance
☐ Yes, parent’s employer insurance	☐ Yes, employer insurance (self)

At the end of the questionnaire, the online patient enrollment system will allow you to upload additional image files. Some examples of documents which may be useful to researchers can include:

Genetic Testing Reports

Values of available labs (most recent):

o Alpha-fetoprotein (AFP)
o CBC – White Blood Cell (WBC), Hemoglobin (Hgb), Hematocrit (Hct), Platelets (Plt)
o Hepatic panel - Total Bilirubin (Tbil), Direct Bilirubin (Dbil), Conjugated Bilirubin (Bc), Alkaline Phosphatase (Alk0), AST, ALT, Gamma-glutamyl transferase (GGT)
o Metabolic panel – Sodium (NA), Creatinine (Cr), Albumin (Alb), Calcium (Ca), Magnesium (Mg), Phosphorus (Phos)
o PT/INR
o Serum bile acids
o Vitamin levels A, D, E

Relevant labs if patient is post-transplant (most recent):

o Hepatic panel - Total Bilirubin (Tbil), Direct Bilirubin (Dbil), Conjugated Bilirubin (Bc), Alkaline Phosphatase (Alk0), AST, ALT, Gamma-glutamyl transferase (GGT)
o Serum bile acids
o tacrolimus levels

Mail-based participants, please include any reports with your questionnaire if you would like to share them with researchers. A CoRDS representative will be able to upload those reports to your account.