

PFIC Network Patient Registry (PNPR): Initiation and Early Analyses

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Background

- Progressive familial intrahepatic cholestasis (PFIC) is an overarching term for monogenic defects in bile function
- PFIC Network is a US-based non-profit patient advocacy organization that aims to improve the lives of affected patient families (www.pfic.org)

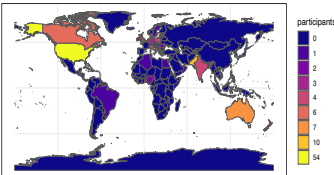
Aims

- To establish a global prospective longitudinal data collection specifically for PFIC and PFIC-related diseases
- To focus on patient-centered outcomes regarding aspects of quality of life in addition to diagnosis, symptoms, medications, and surgeries

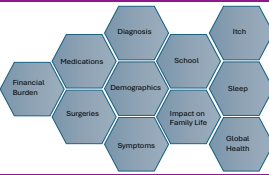
Methods and Results

- Registry data are collected and managed on the REDCap platform and stored on a secure server
- Registry policies and data collection elements are reviewed annually by North Star Review Board, an independent non-profit IRB
- Initial enrollment started in 2022, and participants are invited to fill out a set of follow-up questions every 6 months

Overall Enrollment	127
Sex	
Female	66
Male	60
Prefer not to say	1
Age	
0-5 years	54
6-10 years	20
11-18 years	26
Older than 18	27
Participation in Follow-up waves	
Wave 2	65 (57.5%)
Wave 3	37 (37.8%)
Wave 4	31 (42.5%)
Wave 5	25 (52.1%)



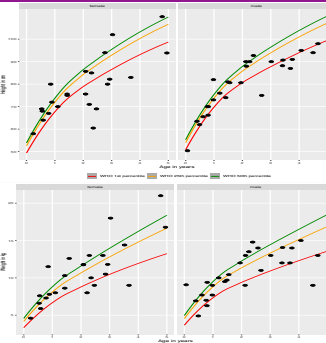
Data collection domains in the PNPR



Medical Data Elements

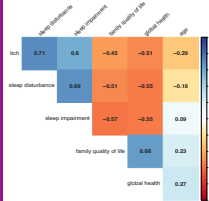
- Diagnosis**
- FIC1: 29
 - BSEP: 40
 - MDR3: 21
 - TJP2: 8
 - MYO5B: 5
 - BRIC: 5
 - Unknown or N/A: 18
- Surgeries**
- Liver Transplant: 27
 - PiBD: 13
 - PEBD: 16
 - Ileal Exclusion: 3
- Reason Liver Transplant:**
- End-stage liver disease: 55%
 - Intractable Pruritus: 38%
 - Liver cancer: 7%

Height and weight of 0-5 year old participants compared to WHO data



Note: The green, orange, and red lines represent the median, the lower 25th percentile, and the lowest 1st percentile of the WHO data. Percentiles can be interpreted as expected percentages, i.e., only 25% of the data should be on or below the orange line, and 1% on or below the red line.

Correlations of itch with sleep, quality of life, and global health



Note: Sleep disturbance measures sleep quality whereas sleep impairment focuses on the consequences of sleep quality.

Data Elements related to Quality of Life

- Itch, sleep, and global health are measured with age-appropriate PROMIS scales.
- Global health includes questions about physical and mental health as well as social functioning.
- PROMIS (Patient-Reported Outcomes Measurement Information System) is a set of person-centered measures that evaluates and monitors physical, mental, and social health in adults and children.
- PROMIS scales are developed and validated according to state-of-the-art psychometric methods.
- Family quality of life is measured with the Impact on Family Scale Revisited developed by Stein and colleagues.

Data Access

- Researchers interested in accessing de-identified registry data can submit a written request to PFIC Network.
- Requests must include an IRB approved research plan along with a form submission. All requests are reviewed by an internal PFIC Network committee consisting of at least two researchers or clinicians and two patients or caregivers.

Financial Burden of PFIC

The PNPR also collects data related to the financial burden of living with the disease. The registry includes 7 questions that were selected from the National Survey of Children's Health (NSCH). The table below shows the results related to three of these questions.

1. During the past 12 months, how much money did you pay out-of-pocket for the medical care of this child? Do not include costs that were or will be reimbursed.	\$0-\$250	\$250-\$500	\$501-\$999	\$1,000-\$5,000	> \$5,000
PFIC families (registry)	25.8	14.5	16.1	24.2	19.4
PFIC families (US only)	23.1	15.4	15.4	23.1	23.5
US general population	60.5	14.2	10.8	12.0	2.5
2. During the past 12 months, have you or other family members left a job, taken a leave of absence, or cut down on the hours you work because of this child's health?	Yes	No			
PFIC families (registry)	64.5	35.5			
PFIC families (US only)	69.2	30.8			
US general population	7.4	92.6			
3. During the past 12 months, have you or other family members avoided changing jobs because of concerns about maintaining health insurance for this child?	Yes	No			
PFIC families (registry)	53.2	46.8			
PFIC families (US only)	57.7	42.3			
US general population	6.6	93.4			

Conclusion

- PFIC is a rare—but often severe—cause of cholestasis that can require medical or surgical intervention and possible liver transplantation
- The goal of the PNPR is to improve our understanding of PFIC diseases, and through that understanding improve the lives of affected patients and their families
- Please contact the PI of the registry, Melissa Kochanowsky at melissa@pfic.org for more information, data access, or to establish contact with participants who opted to be informed about research opportunities

References

- PA Harris, R Taylor, BL Minor, V Elliott, M Fernandez, L O'Neal, L McLeod, G Delacqua, F Delacqua, J Kirby, SN Duda, REDCap Consortium, **The REDCap consortium: Building an international community of software partners**, *J Biomed Inform.* 2019 May 9 [doi: 10.1016/j.jbi.2019.103208].
- Cella, D., Riley, W., Stone, A. A., Rothrock, N., Reeve, B. B., Yount, S., Amtmann, D., D., B., Choi, S., Cook, K. F., et al. (2010). **The Patient Reported Outcomes Measurement Information System (PROMIS) developed and tested its first wave of adult self-reported health outcome item banks: 2005-2008**. *Journal of Clinical Epidemiology* 63, 1179-1194.

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