

# PFIC Network Patient Registry

## Data Summaries - April 2023



### Age by Gender

AGE	[0-1]	[1-3]	[3-5]	[5-10]	[10-14]	[14-18]
FEMALE	6	11	4	4	5	3
MALE	9	5	5	4	3	5

### Subtype by Gender

	TYPE 1	TYPE 2	TYPE 3	TYPE 4	MYO5B	FXR	BRIC	UNKNOWN
FEMALE	9	11	4	2	0	0	1	5
MALE	7	13	2	2	2	0	0	4

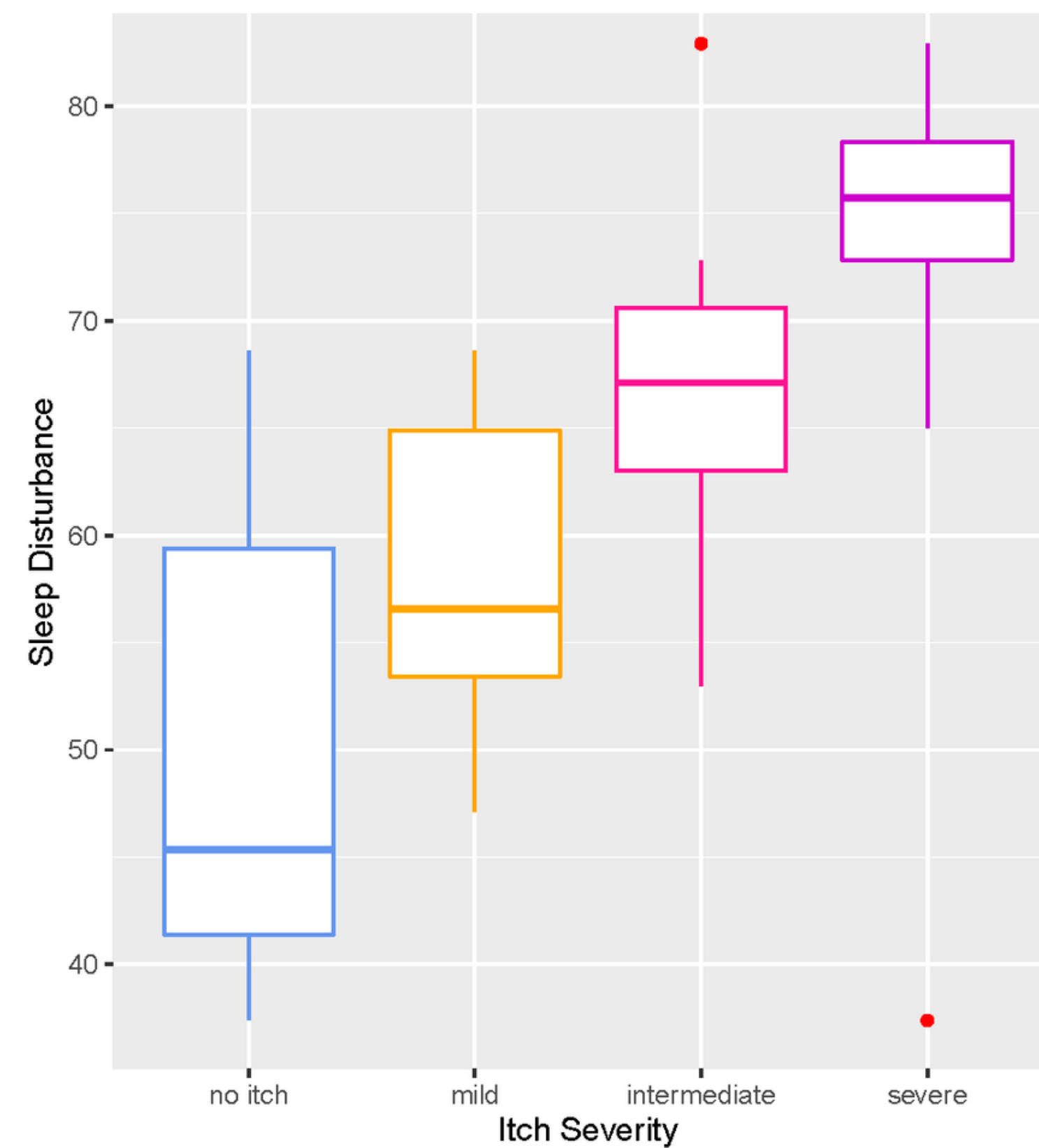
### Surgical Intervention by Subtype

	Partial Internal Biliary Diversion N=7, Age = 4	Partial External Biliary Diversion N=12, Age = 1	Ileal Exclusion N=2, Age = 5	Cholecysto-Appendicostomy N=2, Age = 15	Liver Transplant N=15, Age = 2.5
Type 1 (FIC1, ATP8B1) N=16	3	4	2	2	6
Type 2 (BSEP, ABCB11) N=24	3	5	-	-	5
Type 3 (MDR3, ABCB4) N=6	1	-	-	-	1
Type 4 (TJP2) N=4	-	2	-	-	2
Type Unknown N=9	-	1	-	-	1

This table displays the number of currently enrolled PFIC Network Patient Registry participants by subtype and surgeries received. [N] is the total number of registry participants who indicated the respective variable (e.g. patients with FIC1). [Age] is the median age of the registry participants who indicated that variable (e.g. the median age of patients who indicated they have received liver transplant is 2.5 years old). While the overall number of registry participants is greater, subtype-specific participation numbers are limited. Stand up for your subtype in the PFIC Network Patient Registry so we can learn more about the disease!

(Graph Right) Of the Registry participants that have received liver transplant, 8 participants indicated end-stage liver failure as the reason for receiving transplant. 75% of these participants were between ages 1 and 3 when they received transplant, with a median age of 1, and an outlier at age 10 (red dot). An outlier is a data point that differs substantially from most other data points. 6 participants indicated intractable pruritus as the reason for receiving transplant. 75% of patients with intractable pruritus as their reason received liver transplant between ages 2 and 13, with a median age of 4. The data seems to suggest that both end stage liver failure and intractable pruritus are the more common reasons for liver transplant.

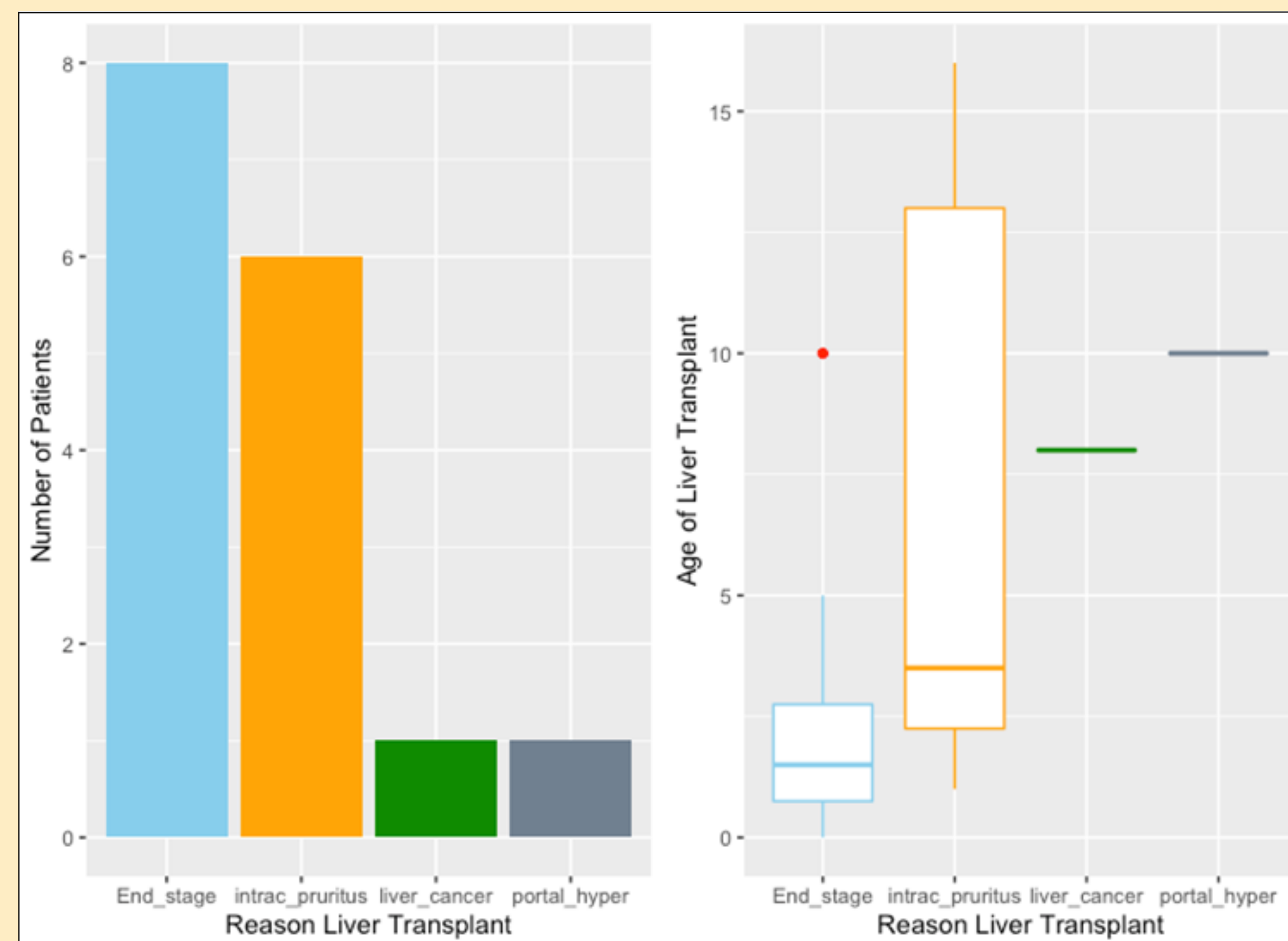
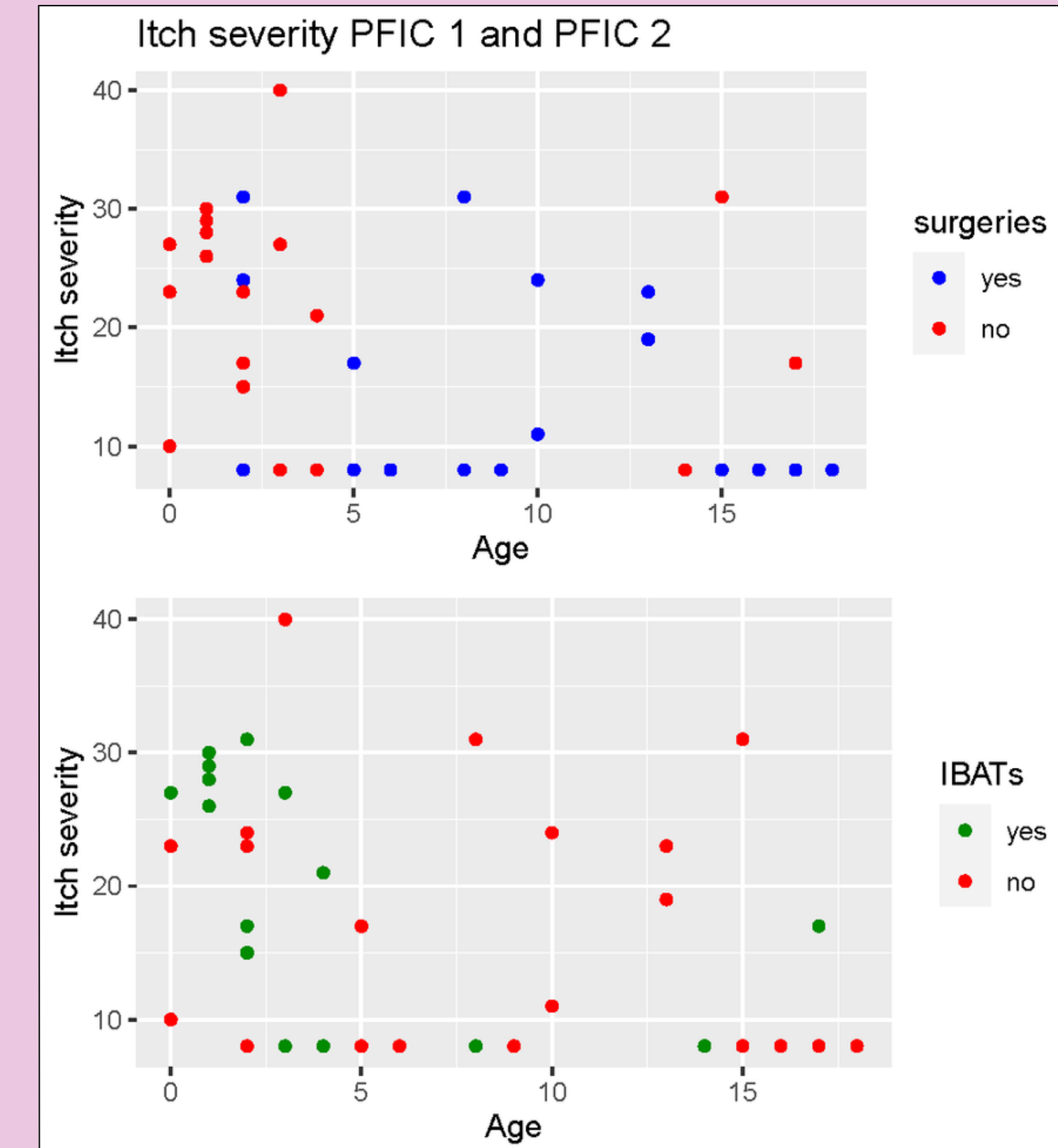
The data from the PFIC Network Patient Registry presented in the graph to the right suggests a relation between itch severity and amount of sleep disturbance. Itching can severely disrupt sleep, and not sleeping might also make the disease worse and increase itch. This relationship between itch and sleep can feel like a no-brainer for our community. We know the struggle very deeply. But it is important to give evidence to this experience through data, so that researchers can be alerted to the impact of PFIC on quality of life and the severity of that impact. This type of relationship has not been researched very much in PFIC. By having greater participation in the PFIC Network Patient Registry, we can collect more data on important variables like itch and sleep that can greatly inform PFIC research.



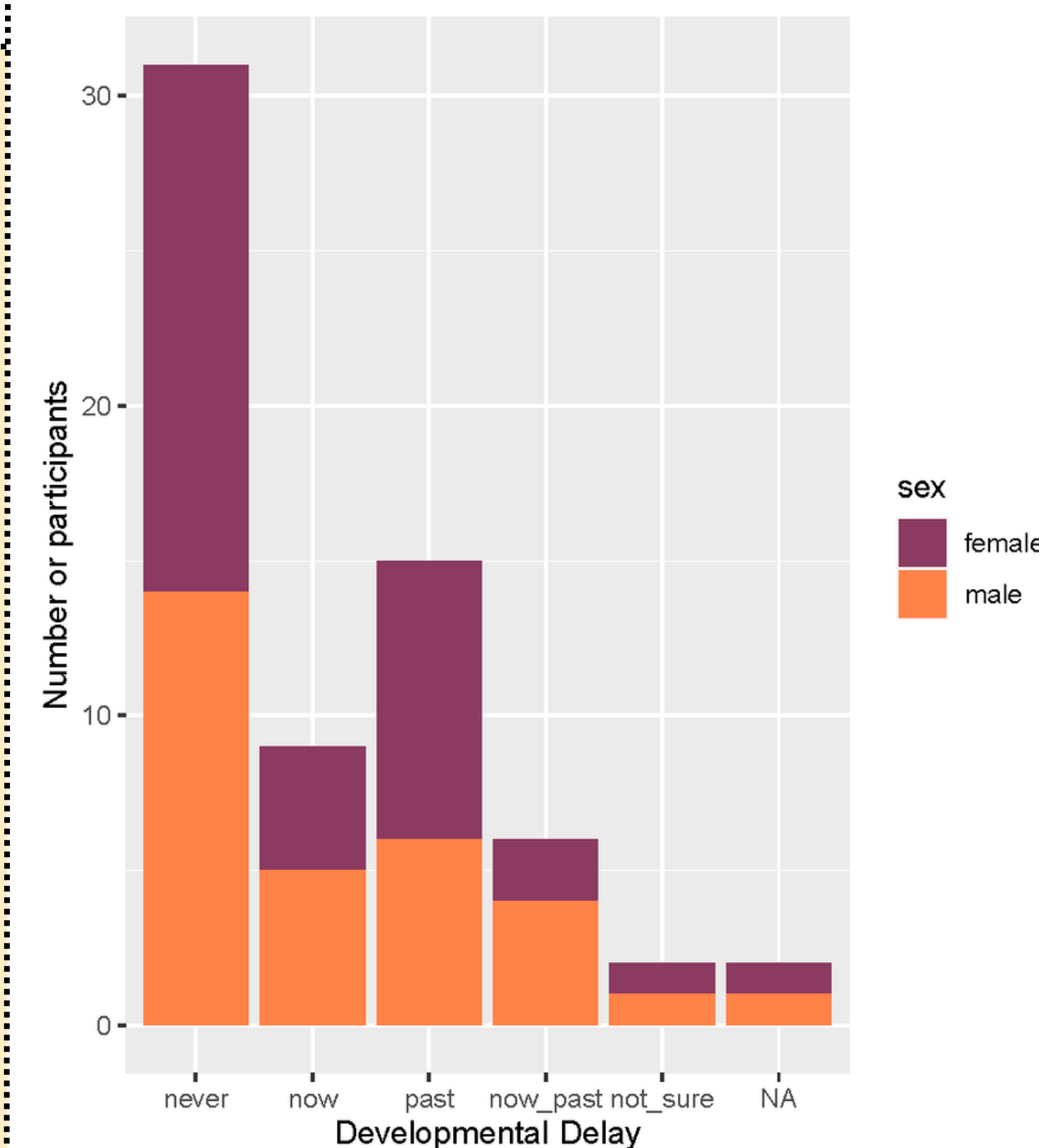
In the two graphs on the right, you can see that PFIC 1 and PFIC 2 participants in the registry who reported low itch severity have either had surgery (blue dots in upper plot) or take IBATs (green dots in lower plot). There are currently no patients in the registry who have reported experiencing low itch severity and have not received surgery or medical treatment.

What does this tell us? PFIC patients intervene for itch, but itch can remain a problem even after intervention.

Of course it is possible that patients on IBATs or after surgeries are still itching because they just started their treatment or had their surgery. That is why follow-up data are so important!



Please note: these participation numbers are low and may not represent all PFIC patients. The greater the registry participation, the more reliable the data can become!



Almost half of the participants in the PFIC Network Patient Registry have indicated experiencing some developmental delays. The data below also suggests that there is no difference between the sexes in their experience of developmental delays now or in the past. In other words, the data suggests that gender has no bearing on likelihood of experiencing developmental delay. But please remember: these participation numbers are low and may not represent all PFIC patients. By having greater participation in the PFIC Network Patient Registry, we can learn a lot of important information about PFIC – such as its effect on factors like development and sleep!