PFIC IS A GROWING GROUP OF DISORDERS, WITH MANY DISTINCT SUBTYPES THAT PRESENT DIFFERENTLY[1-5]

Mutated gene	ATP8B1	ABCB11	ABCB4	TJP2	NR1H4	SLC51A	USP53	KIF12	ZFYVE19	MYO5B	SEMA7A	VPS33B	PSKH1
Protein deficiency	FIC1	BSEP	MDR3	TJP2	FXR	OSTα-OSTß	USP53	KIF12	ZFYVE19	MYO5B	SEMA7A	VPS33B	Putative protein serine kinase of unknown function
Age of onset	0-3 months	0-4 months	2-3 years	0-3 months	0-3 weeks	0-3 days	0-15 years	0-4 weeks	0-9 years	0-2 years	0-6 months	0-4 weeks	Typically, infancy or first years of life, although some present later
Pruritus intensity	Severe	Severe	Severe	—	—	—	Mild	—	—	Moderate	None	Severe	—
Bile acid level	High	High	Normal	High	High	Normal	High	High	High	High	High	High	Elevated
Transaminases (AST and ALT)	—	Elevated	—	—	—	—	—	—	—	—	—	—	Elevated
GGT	Normal	Normal	Elevated	Normal	Normal	Elevated	Normal	Elevated	Elevated	Normal	Normal	Normal	—
Other features	Hearing loss	Risk of liver cancer	Risk of liver cancer	Hearing loss	Coagulation problems	Elevated cholestasis	Hearing loss	Neonatal cholestasis	Gastrointestinal bleeding and diarrhea	Pyramidal syndrome	Jaundice	Intense pruritus	—

These data were compiled from multiple sources.

ALT=alanine aminotransferase; AST=aspartate aminotransferase; GGT=gamma-glutamyl transferase; PFIC=progressive familial intrahepatic cholestasis.

Dashes (—) indicate the feature is not applicable, not observed, or no specific data is available for that category.

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