# PFIC Fact Sheet

## Progressive Familial Intrahepatic Cholestasis

### WHAT IS PFIC?

Progressive familial intrahepatic cholestasis (PFIC) is a spectrum[1-4] of autosomal recessive genetic disorders in which cholestasis leads to liver failure.[5]

The estimated incidence of PFIC ranges from 1 in 50,000 to 100,000 births.[5] Subtypes PFIC1, PFIC2 and PFIC3 are most common.[5] In addition, other rare forms of PFIC exist with varying phenotypes, but all present with cholestasis.[6]

The most debilitating symptom of PFIC is pruritus (insatiable itching), which may be so severe that it leads to skin mutilation, loss of sleep, irritability, poor attention and impaired school performance.[9] Up to 80% of PFIC patients had pruritus graded as severe (associated with abrasions, skin mutilation, hemorrhage or scarring).[10]

Survival analysis showed that at 18 years of age, only 44% of PFIC1 patients and 32% of PFIC2 patients were alive with their native liver.[7,8]

Pruritus is a primary cause of surgical treatments and transplant, cited as the indication for surgical diversion in the majority of PFIC patients[7,12] and for transplant in 50% of patients with PFIC1.[13]

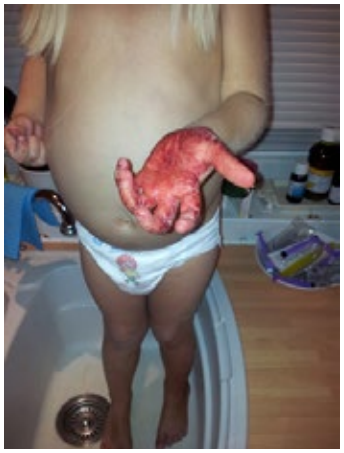
### SIGNS AND SYMPTOMS OF PFIC

The signs and symptoms of PFIC usually present in infancy in PFIC1 and PFIC2 and later in infancy to young adulthood in PFIC3.[9,11] They may include:

- **Pruritus**
  - Insatiable itching that may lead to skin mutilation, loss of sleep, irritability, poor attention and impaired school performance[9]
- **Hepatic manifestations**
  - Jaundice (yellowing of the skin and whites of the eyes)[9]
  - Elevated serum bile acids[11]
  - Hepatomegaly (enlarged liver)[9]
  - Cirrhosis and end-stage liver disease[11]
  - Increased risk of liver cancer (PFIC2)[9]
- **Gastrointestinal symptoms**
  - Gallstones[11]
  - Pancreatitis (PFIC1)[11]
  - Fat malabsorption[9]
  - Diarrhea[10]
  - Pale/discolored stools[10]
- **Metabolic defects**
  - Fat-soluble vitamin (A, D, E and K) deficiency[9]
  - Growth retardation[9]
  - Bleeding (due to vitamin K deficiency)[11]

## DIAGNOSIS OF PFIC

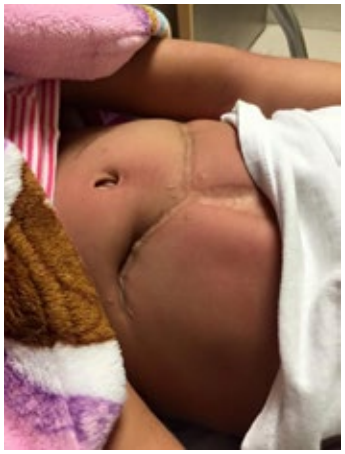
Diagnosis should utilize a combined clinical, biochemical, radiological, and histological approach.[5] In order to diagnose PFIC and the specific subtype, testing may include liver function tests, liver ultrasound and biopsy, immunohistochemistry, bile analysis, and genetic testing.[9]



## SURGICAL TREATMENTS FOR PFIC

Surgical treatment options for PFIC include biliary diversion procedures and liver transplantation.[9]

PEBD decreases bile through ostomy.[11] Ostomy requires maintenance and induces fluid/electrolyte loss.[11,14] In a study of 33 patients with cholestatic liver disease who had surgical biliary diversion, stoma complications occurred in 55% and 20 secondary surgeries were required.[12]



Patients undergoing liver transplant require long-term management with immunosuppressive medication, which can increase the risk of metabolic disorders such as diabetes, hypertension, hyperlipidemia, osteoporosis and chronic kidney disease.[15] Nearly a quarter of liver transplants in children fail within the first 6 months, almost a third within 5 years and almost half within 20 years.[16]

## RESOURCES FOR MORE INFORMATION

For information about treatments, talk to your doctor, and visit **AlbireoAssist.com**.

For more information on living with PFIC, visit:

PFIC Voices: **PFICVoices.com**

National Institutes of Health: **https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases**

PFIC Advocacy and Resource Network: **https://www.pfic.org/**

## REFERENCES

1. Henkel S et al. World J Hepatol. 2019;11(5):450-463. 2. Schatz BA et al. Hepatol Commun. 2018;2(5):504-514. 3. Aldrian D et al. J Clin Med. 2021;10(3):481. 4. Folmer DE et al. Hepatology. 2009;50(5):1597-1605. 5. Davit-Spraul A et al. Orphanet J Rare Dis. 2009;4:1. 6. Amirneni S et al. World J Gastroenterol. 2020;26(47):7470-7484. 7. van Wessel D et al. Hepatology. 2021;10.1002/hep.31787. 8. van Wessel D et al. J Hepatol. 2020;73(1):84-93. 9. Srivastava A. J Clin Exp Hepatol 2014;4:25-36. 10. Baker A et al. Clin Res Hepatol Gastroenterol. 2019;43(1):20-36. 11. Gunaydin M et al. Hepat Med. 2018;10:95-104. 12. Bjørnland K et al. Eur J Pediatr Surg. 2020. https://pubmed.ncbi.nlm.nih.gov/32707578/ 13. van Wessel et al. Supp to: Hepatology. 2021. 14. Stapelbroek JM et al. J Hepatol. 2010;52(2):258-271. 15. Singh S et al. Mayo Clin Proc. 2012;87:779-790. 16. Venick et al. J Am Coll Surg. 2018;226(4):355-366

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