



Talking with your Doctor

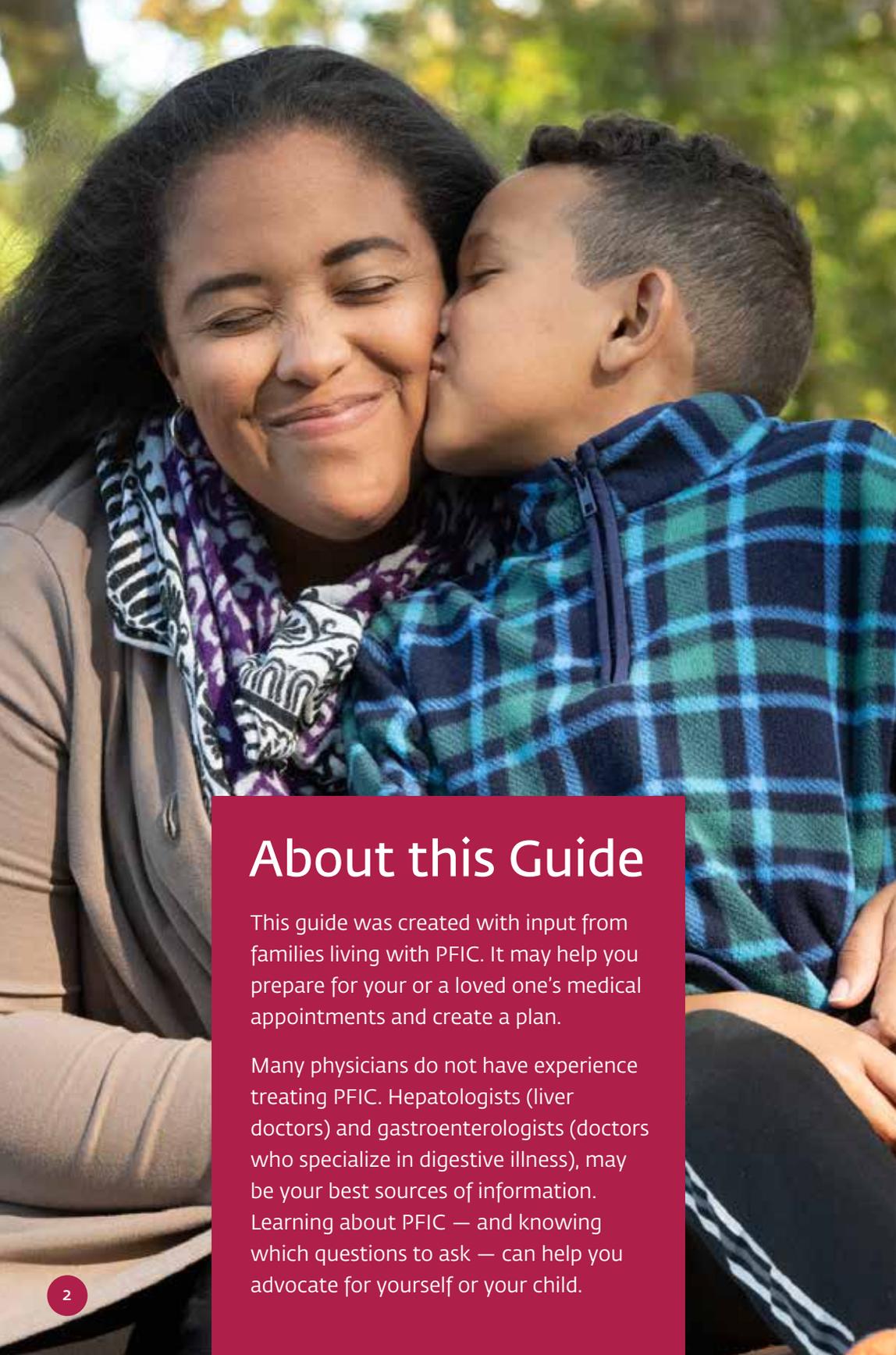


PFIC VOICES[®]

Progressive Familial
Intrahepatic Cholestasis

Created in partnership with PFIC Network





About this Guide

This guide was created with input from families living with PFIC. It may help you prepare for your or a loved one's medical appointments and create a plan.

Many physicians do not have experience treating PFIC. Hepatologists (liver doctors) and gastroenterologists (doctors who specialize in digestive illness), may be your best sources of information. Learning about PFIC — and knowing which questions to ask — can help you advocate for yourself or your child.

Talking to Your Doctor About PFIC

Progressive familial intrahepatic cholestasis (PFIC) is a spectrum of rare, inherited liver diseases that can take a devastating toll on patients and their families. It can lead to severe itching (pruritus), slowed growth, and liver failure.

The itching can be so severe that children and infants may scratch through their skin.¹ Many patients turn to surgery, including liver transplant, to relieve the itching.^{2,3} Itching can also cause loss of sleep, poor attention, and lower school performance.¹

PFIC Signs and Symptoms

- Severe itching (pruritus)¹
- Yellowing of the skin (jaundice)¹
- Problems absorbing fats and fat-soluble vitamins (vitamins A, D, E and K)¹
- Problems with bile flow that can lead to liver failure⁴

Normally, the body makes bile acids that help with digestion, including absorbing fats and some vitamins. In people with PFIC, the normal flow of bile acids is disrupted. Bile acids build up in the body, damaging the liver.^{6,7}

PFIC affects 1 in 50,000 to 100,000 births.⁴ It is caused by mutations on different genes, including ATP8B1 (PFIC1), ABCB11 (PFIC2), ABCB4 (PFIC3), and others.⁵ PFIC1, PFIC2, and PFIC3 are the most common types.⁴ In addition, other rare forms of PFIC exist. They have some differences, but all cause problems with bile flow.⁵ Genetic testing can be relied on to support but not always definitively confirm the clinical diagnosis of PFIC.⁸

Easy Ways to Prepare for a Doctor Visit

Planning ahead for your doctor's visit can help you get the most out of each appointment. Here are steps you can take to prepare.



List your questions in advance.

During the course of a normal day, you may think of questions to ask your doctor. It can be easy to forget what you wanted to ask on the day of your actual appointment. Keeping an ongoing list of questions on your phone can help you remember important topics.



Bring a notebook, and maybe a companion.

Your healthcare provider may cover many topics. Taking notes can help ensure that you have a record of what you talked about. You can also ask the doctor or nurse to write down important points or print out a copy of the visit's notes. Also, it can be helpful to have someone with you to take notes and help you remember everything you wanted to talk about. If you have a young child, a companion can sit with your child while you're talking with the doctor.



Keep track of medications and dosages.

Tell your doctor about any medication or topical treatment being taken. Be sure to track dosages, how helpful the medications have been and side effects experienced.



Keep a journal and bring it to your appointment.

A written record can help you share symptoms and concerns. For instance, if you or your child often lose sleep because of severe itching, you may want to track sleeping patterns to discuss possible solutions with your doctor. You can also write down what you or your child eats to help your doctor address any nutritional needs.



Bring activities.

If you are taking a younger child to the doctor, a toy or game can help pass the time when you are waiting for the appointment to start.



Explain to your child what to expect.

Doctor's appointments can be a scary experience for young children. Help them prepare for the appointment by sharing what you know in a way that they may be able to understand.

During Your Appointment



Ask your most important questions first.

If you run out of time, you'll have the answers you need most.



Ask more questions.

If you don't understand something your doctor says, ask for a better explanation. Try to make sure that you leave the appointment feeling comfortable that all your questions have been answered. There is never a wrong question. Ask all questions that come to mind, even if you feel like you might know the answer.

Questions to Ask Your Provider

PFIC is a progressive condition, which means that it may get worse over time. Keeping a log of questions to ask at every appointment may be helpful for tracking disease changes or progression.

About PFIC Symptoms and Management

- What can I expect from PFIC? What risks and symptoms should I know about?
- What are the options for medication and how can they help? What side effects can I expect from the medications?
- Other than medication, what can I do to reduce symptoms?
- How will I know if and when my child needs a surgical procedure or transplant?
- Should my child get a genetic test for PFIC?
- Are there any resources that you can recommend that provide helpful information about PFIC and its symptoms?
- Is my child receiving adequate nutrition? What are signs or symptoms that we should be aware of to monitor vitamins?



About Your Appointments

- How often should I expect appointments? What does each appointment involve?
- What tests should I prepare for? How long will they take?
- When will I have my child's results?
- How can I keep track of my child's test results?
- If I have a concern about my child's symptoms, who should I contact?

Other Healthcare Needs

- Can you help me find a support group?
- Is all testing, including a genetic test, covered by insurance?
- How can living with PFIC affect my child's mental health? Do you have a psychologist you recommend if we need one?



Remember

Your doctor is a member of your healthcare team. Open communication will help build trust so you can achieve the best outcomes for your family.

This brochure is published by PFIC Voices (an initiative of Albireo Pharma) in partnership with the PFIC Network, a parent- and patient-led advocacy organization.

About Albireo

Albireo is committed to supporting people with PFIC, their families, and healthcare providers with resources to better understand and manage this devastating disease. The company is focused on the development of novel bile acid modulators to treat rare pediatric liver diseases, such as PFIC.

For more information and resources about Albireo's work with the PFIC Community, visit www.PFICvoices.com.

References

1. Srivastava A. J Clin Exp Hepatol. 2014;4:25-36.
2. Bjørnland K et al. Eur J Pediatr Surg. 2020.
3. van Wessel DBE et al. Hepatology. 2021;10.1002/hep.31787.
4. Davit-Spraul A et al. Orphanet J Rare Dis. 2009;4:1.
5. Amirneni S et al. World J Gastroenterol. 2020;26(47):7470-7484.
6. Kamath BM et al. Liver Int. 2020;40(8):1812-1822.
7. Karpen SJ. Clin Liver Dis. 2020;15(3):115-119.
8. Bull LN et al. Clin Liver Dis. 2018;22:657-669.

