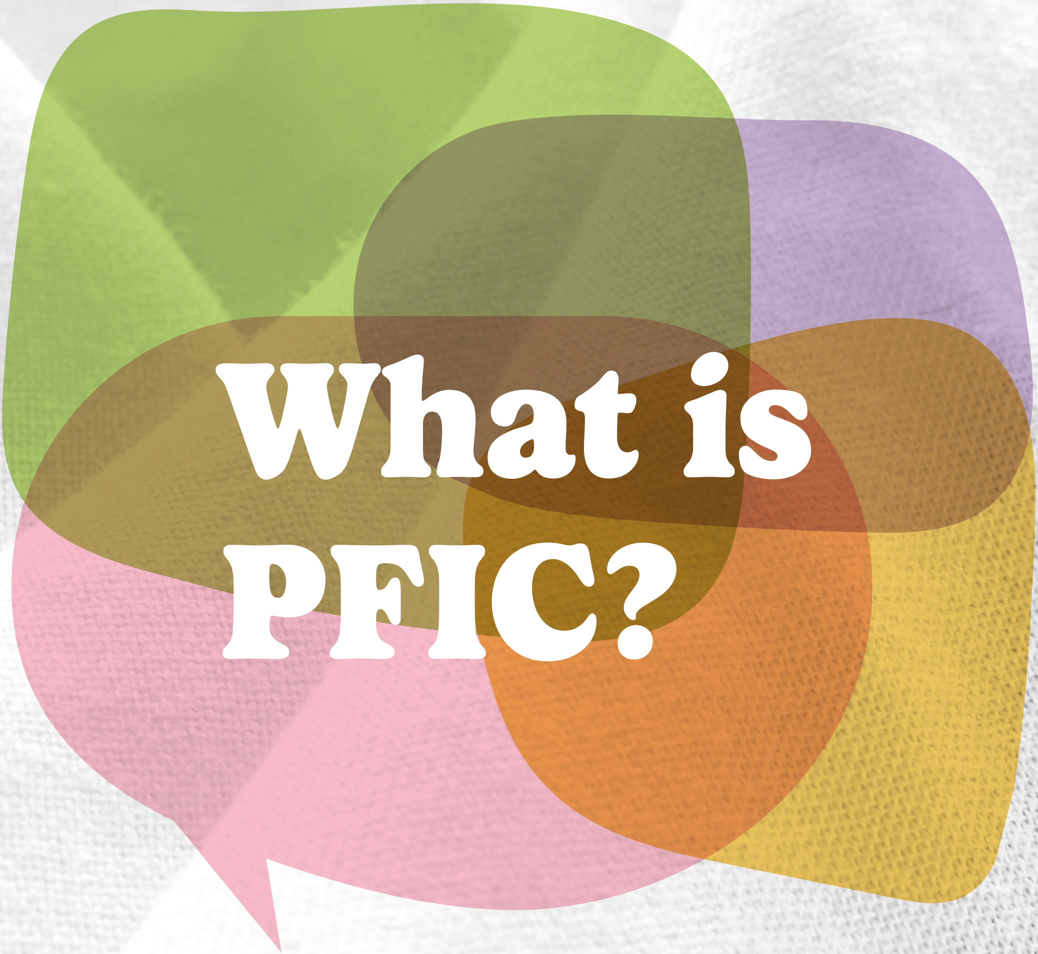

What is PFIC?

A guide for you and your loved ones.

This brochure has been designed for patients, parents, caregivers and the families of children diagnosed with progressive familial intrahepatic cholestasis (PFIC).

You are not on your own.

This brochure was commissioned by Ipsen and has been developed with input from liver specialists.

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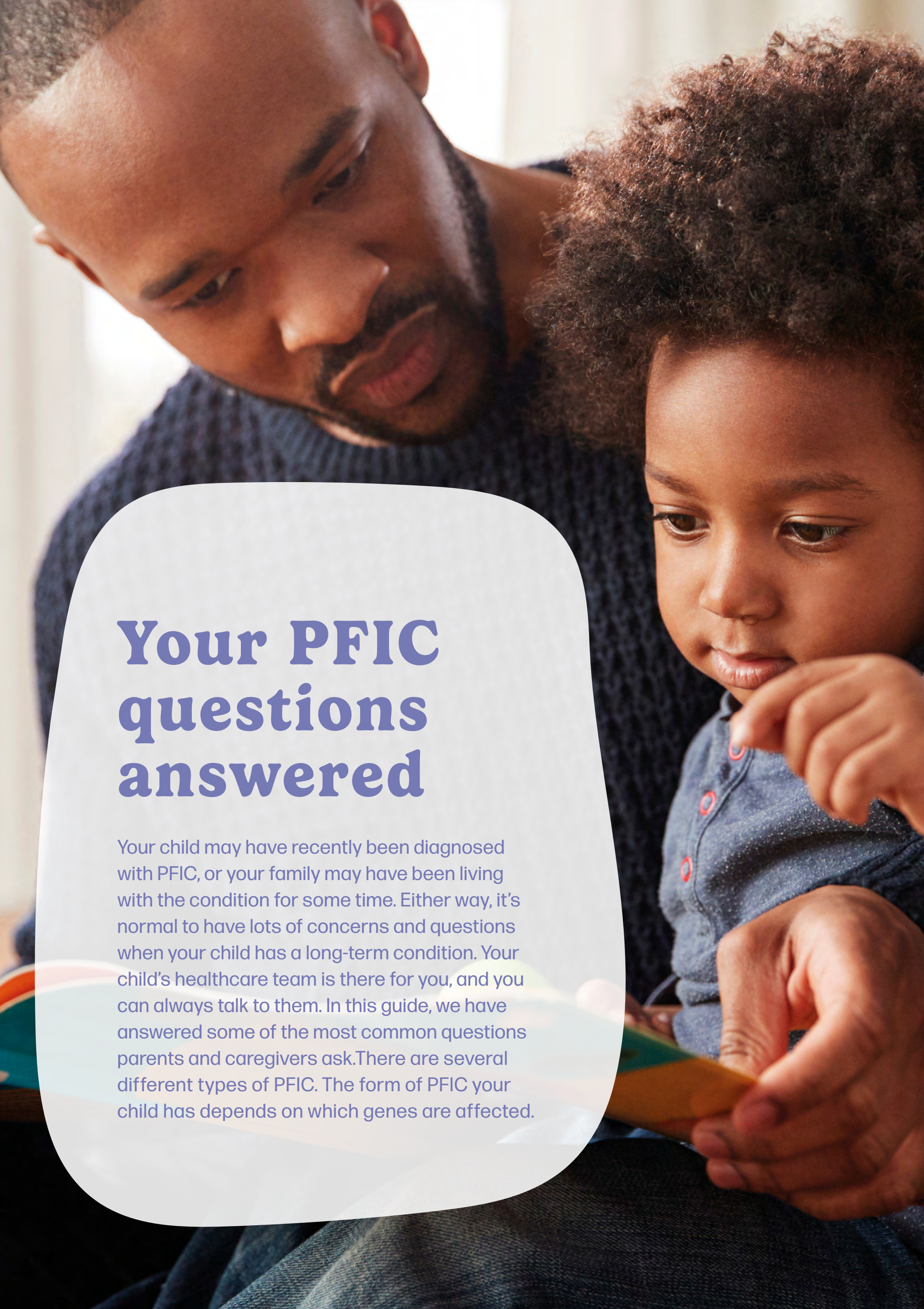

Your PFIC questions answered

Your child may have recently been diagnosed with PFIC, or your family may have been living with the condition for some time. Either way, it's normal to have lots of concerns and questions when your child has a long-term condition. Your child's healthcare team is there for you, and you can always talk to them. In this guide, we have answered some of the most common questions parents and caregivers ask.There are several different types of PFIC. The form of PFIC your child has depends on which genes are affected.

What is PFIC?

PFIC is a group of genetic conditions which stop the liver from properly releasing a digestive fluid called bile. Normally, bile is made in the liver and stored in the gallbladder. It's then released from the gallbladder by bile ducts and taken down into the intestine.

In the intestine, bile helps the body digest food. In people who have PFIC, the bile isn't released and it builds up in the liver instead. The build-up of bile is called cholestasis. This can start to damage the liver and cause scarring, known as cirrhosis.

There are several different types of PFIC, depending on which genes are affected. If you have any questions about the type of PFIC your child has, speak to your specialist doctor or nurse.

Key language

Progressive: likely to get worse over time

Familial: linked to genetics in families

Intrahepatic: a disease inside the liver

Cholestasis: poor flow of bile, and build-up of substances in the liver

What is bile and why is it so important?

Bile is a green-yellow fluid made in the liver. It has important functions, including breaking down fats so they can be absorbed, helping the body to absorb certain vitamins and removing waste products. It's made up of different substances, including:

- bilirubin (a waste product formed from old red blood cells, which gives bile its distinctive colour)
- bile acids (which help to digest fats)
- cholesterol
- fats
- water
- other waste products

Why does my child have PFIC?

Our body's cells make proteins that do different jobs in the body. These proteins are made using instructions from a pair of matching genes. We inherit one gene from our mother and one from our father. Sometimes there's a mistake in a gene, known as a mutation. If a child inherits one healthy gene and one mutated gene, the healthy one can usually make up for the mutation so it doesn't cause problems in their body. But if a child inherits a mutated gene from each parent, it can result in a genetic condition, like PFIC.

How is PFIC treated?

Your child may need special foods and supplements to make sure they're getting enough nutrients. Sometimes, a child may be given food through a nasogastric tube which goes through a nostril and into the stomach. Your doctor will advise you of other management options available for your child.

What will happen to my child in the future?

PFIC progresses differently in different children. Some children will need a liver transplant, either because their liver has become too damaged or because their itching is unmanageable.

Your child's healthcare team can provide more information about liver transplants and available management options.

What symptoms does PFIC cause?

As the liver becomes damaged it causes a number of symptoms, including the following:

- **Jaundice.** This is when the skin and whites of the eyes turn yellow. It happens because dark-coloured bilirubin passes back into the bloodstream and travels around the body. It can also cause developing teeth to become discoloured and lead to dark urine.
- **Pale, greasy poo.** You may find it floats in the toilet. This is because your child's poo contains high levels of undigested fat, as there isn't enough bile going from their liver to the intestine to help with digestion.
- **A swollen tummy.** Your child might experience a 'full' feeling or discomfort in their abdomen (tummy). This can be caused by swelling in their liver.
- **Vitamin deficiencies.** This happens because the body doesn't absorb nutrients properly. Your child is especially likely to lack the fat-soluble vitamins, A, D, E, and K. These are needed for different functions in the body, including healthy bones, eyesight, and the nervous system.
- **Slower growth.** This means your child doesn't grow as expected and happens because they don't absorb fats.
- **Itching.** This can often be the most difficult symptom to manage. The cause is not fully understood. On the next three pages, you can read more about itching and get tips from other parents on how to manage it.

PFIC itching

Parents and patients usually describe this as the most disruptive symptom of PFIC as it can affect the whole family.

Children often cannot resist scratching, which can lead to bleeding and scarring. Itching can disrupt sleep, and impact day-to-day life and school attendance.

Itching is a symptom that may not be understood by everyone. This myth-buster looks at some of the common misunderstandings, gives you the facts, and suggests some tips that may help.

> “Itching isn’t that bad”

People may not understand why itching can be so difficult to deal with

The PFIC itch is completely different to ‘normal’ itching.

Most people think the itching must be the same kind of itching you get from insect bites or dry skin conditions. It can be very frustrating when others think like this. As PFIC is a rare disease, some healthcare professionals may not understand this well either.

It’s often described as an intense ‘internal itch’, which the child attempts to soothe by scratching their skin. This may give some temporary relief, before becoming worse again. The itch is almost always present. It can be better or worse at different times, but it doesn’t necessarily go away completely.

Babies can’t scratch themselves, so the earliest sign may be poor sleep and irritability. Older children may make themselves bleed by scratching.

Itching may affect your child’s experience at school

Some children with PFIC struggle at school because they miss out on sleep. They may have trouble focusing and need lots of time off. Some may have some developmental delays because they can’t engage with normal activities.

PFIC can make children look different, for example, their skin may be yellow from jaundice, their stomach may be swollen, and they may have scabs and scars from scratching.

Ways to support your child

Parents say it’s important to help your child feel proud of themselves, whatever their skin looks like.

You may find that talking to other families who are living with PFIC can help support your family.

Talking to your child’s teachers about their condition can help everybody understand what your child may be struggling with and any differences in their appearance.

Because the itch can be so difficult to live with, in some cases your healthcare team may recommend a liver transplant.

> “At least it isn’t life-threatening”

What helps the PFIC itch?

You may have already found some ways to help your child with the itching. These are ideas that other parents and caregivers have found helpful. Every child is different, so you'll probably need to try a few ideas to see what helps your child. Keep a record of any techniques you try that seem to be effective.

Choose soft fabrics. Parents and caregivers usually say loose cotton clothes are best, and recommend avoiding wool and synthetic fibres, which may make the itching worse.

Keep your child cool. Heat usually makes itching worse so try to keep your home cool. Some parents say they apply soothing lotions straight from the fridge. You could also try giving your child cool baths with some baby lotion added.

Keep your child's nails short so they can't damage their skin too much. You could encourage them to try rubbing or stroking their skin instead of scratching.

For babies and small children, try to dress them in clothes that cover their whole body. You may find sleepsuits with cuffs that fold over their hands and feet helpful. In the day, dungarees may be a good idea.

Other parents and caregivers say distracting their child is helpful. Although you have a lot to think about, you may find that planning activities to do at home, family outings and social opportunities can give your child things to look forward to and help provide a distraction.

Do you have the support you need?

Lots of parents and caregivers of children with PFIC say they find it helpful to have support from friends and family. This can be both practical and emotional support.

It isn't just about direct support with PFIC-related tasks. Anything that gives you a little time to yourself can help you look after your own well-being and energy levels.

You may not feel you need any help, and that's OK, too. We're all different. But sometimes even the occasional bit of extra support can make a big difference.

Simply knowing you can ask for help can be reassuring. Your need for support may change over time, so it's a good idea to have a support network in place in case you need it in the future.

It isn't always easy to ask for help but remember, we all give and receive support at different times. It's likely people in your life want to help you. They may just not know how.

You're not alone

Every child with PFIC is different, and every family's experience is different. Families have told us about some of their challenges.

- Parents and caregivers say they often feel guilty about focusing more on their child with PFIC than their other children.
- Other children may feel guilty for being healthy, or resentful that they don't get as much attention.
- Everyone's sleep may be affected by the child with PFIC waking and crying during the night.
- Parents and caregivers may not have much time or energy for their relationships, which can lead to difficulties.
- Managing your child's symptoms and healthcare appointments may need to take priority over fun and relaxing time.
- A child with PFIC may need hospital stays. This can mean they and at least one parent or caregiver are away from home and the rest of the family for long periods of time.
- It's likely one parent or caregiver may have had to stop working or reduce their hours to care for their child with PFIC. This may lead to money worries in the family.

Families have also told us that they have found some positives in the situation.

- Other children can learn a lot of strength and kindness from helping to look after a brother or sister who is not well.
- With support, a child with PFIC can become resilient and confident.
- Parents and caregivers become advocates for their child. Many learn they have strength they didn't know they had.
- All family members can learn to appreciate simple, everyday happiness.

Live well as a family

Having a child with a long-term condition like PFIC can have a huge effect on everyone in the family. Parents and caregivers often tell us the whole family is affected by PFIC, not just the child with the condition. If your child has only recently been diagnosed with PFIC, you may all be adjusting to the changes. If your family has been living with PFIC for a while, you'll probably know that many aspects of family life can be affected. You may be wondering whether things could be easier.

Supporting your family

1 Find the positives

It's likely you'll have good and bad days with PFIC. There's still room for fun and laughter. Other parents and caregivers tell us they've found it's important to stay united as a family. It may be helpful to sit down together and think about what being a positive family means to you. Some ideas may include:

- being able to talk openly about feelings
- physical touch
- spending time in nature
- sitting down to eat together
- making plans for the future
- supporting each other's choices.

2 Solve problems

Taking a step-by-step approach to solving problems as a family can help you work together. Talk to each other to identify the problem, and the advantages and disadvantages of any solutions you can think of. Agree on one thing to try first. After a while, discuss whether it is helping. If not, you can try another approach.

3 Accept difficult feelings

There are some things you can't change. In these situations, learning to accept your feelings can be helpful. Avoiding or ignoring painful emotions can make you feel worse or lead you to try and cope in unhealthy ways. Talking about your feelings with family also helps your children learn how to deal with bad feelings.

4 Make time for fun

Your child and your family are much more than PFIC. Having fun as a family can improve quality of life and may even be helpful for distracting your child if they're itching. Other parents and caregivers in PFIC support groups can give you some more ideas of what you can do to enjoy time together as a family. These groups may also have family days so you can meet other families living with PFIC for fun activities. Look for support groups on social media or visit childliverdisease.org.

If your brother or sister has PFIC

It can be very difficult having a brother or sister who's not well. You may feel lots of different things and have the following thoughts:

- I feel very sad for my brother or sister when they don't feel well.
- I get annoyed when my brother or sister cries and keeps me awake at night.
- It's not fair that sometimes my parents don't have time for me.
- I wish I could make my brother or sister feel better.
- I want to have a normal family with a brother or sister who isn't ill.

It's normal to have all these sorts of thoughts when you have a brother or sister who's ill. It can help to remember that lots of children have a brother or sister who's ill, and many of them have the same thoughts and feelings.

Sometimes your parents may have to give your brother or sister more attention, but they still love you. It's important to tell them if you're feeling bad. They can make you feel better.

It might also help if you have a teacher or a friend's parent who you can talk to.

You might want to help your parents look after your brother or sister. Playing a game together might stop them thinking about how itchy they feel, or you could help put on some cooling cream that makes them feel better. You can be a very special brother or sister. You might have days you don't want to help and that's OK. It's not easy having a brother or sister who isn't well.

You can ask your parents if you can talk online to other children who have a brother or sister with PFIC. In different countries all over the world there are children who have brothers and sisters with PFIC. You will probably have lots of fun getting to know them and hearing about their lives. You might be able to meet up with them at big family days!

Hope for the future

Having a child with PFIC can lead to a lot of anxieties. But it's important to know that doctors are finding out more about the disease all the time. Your child's healthcare team and other families who are living with PFIC are your best sources of support in finding out about new research and ways to manage the disease from day to day.

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Having a child with a rare condition like PFIC can be isolating. That's why it's so important to connect with other families.

Look for support groups on social media, or visit the website below:

childliverdisease.org

The information in this brochure has come from the following sources:

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