



Children's Liver
Disease Foundation

fighting childhood
liver disease

Fibropolycystic Liver Disease

A Guide



An overview of fibropolycystic liver disease,
including types, causes, diagnosis, symptoms,
complications and treatment

This information has been written for:

- parents and carers of children and young people with fibropolycystic liver disease

Others who may also find this information useful:

- young people with fibropolycystic liver disease
- healthcare professionals who would like to find out more about fibropolycystic liver disease

It provides information on:

- the different types
- causes
- diagnosis
- symptoms
- possible complications
- treatments

You may also find it helpful to read the following CLDF leaflets:

- Introduction to Liver Disease
- Portal Hypertension and Ascites
- Liver Transplantation

Use this
QR Code to view
our resources



Key facts about fibropolycystic liver disease

- 1** Fibropolycystic liver disease is a collective term for a group of rare conditions. These conditions affect the liver and the biliary tract. They may also involve the kidneys.
- 2** The term fibropolycystic liver disease covers the following conditions:
 - Congenital hepatic fibrosis
 - Caroli disease
 - Caroli syndrome
 - Biliary hamartomas
 - Polycystic liver disease
- 3** The conditions are grouped together because they begin in the body in the same way. They all start before birth.
- 4** There is often some overlap between the conditions. Your child may have one condition, or a combination of more than one.
- 5** The symptoms and complications of the different conditions can vary widely.
- 6** The conditions often involve the development of cysts. Cysts are pockets of tissue filled with fluid that develop inside an organ. They can also occur in the bile ducts.
- 7** There will often be problems in the kidneys that go alongside those in the liver.
- 8** Treatments focus on reducing symptoms and complications. There are currently no cures available.
- 9** Your child may see a range of professionals. This may include liver doctors, kidney doctors, specialist nurses, radiologists, and surgeons.
- 10** Support services are available from Children's Liver Disease Foundation.

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What is fibropolycystic liver disease?

Fibropolycystic liver disease is a collective term for a group of rare conditions. These conditions affect the liver and the biliary tract. They may also involve the kidneys.

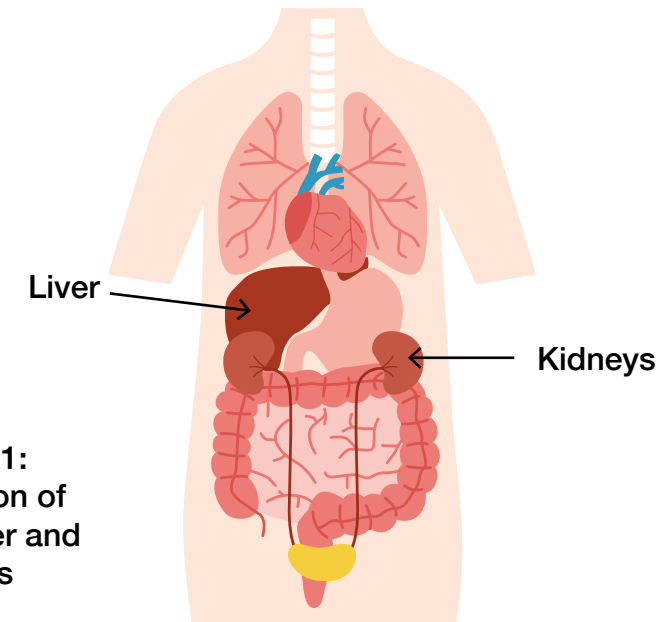


Figure 1:
Location of
the liver and
kidneys

This leaflet covers the following conditions:

- Congenital hepatic fibrosis
- Caroli disease
- Caroli syndrome
- Biliary hamartomas
- Polycystic liver disease

These conditions are grouped together because they all begin in the body in the same way. This also means there is often overlap between them. Your child may have one of these conditions or a combination of more than one.

The conditions are all caused by changes (mutations) in a person's genes. Some genetic changes are 'autosomal recessive'. This happens when both parents are carriers of the change and pass it on to their child. Other genetic changes are 'autosomal dominant'. This means that only one parent carries the change and they pass this on to their child.

Fibropolycystic liver diseases begin before birth. They happen when ductal plates do not form properly during the early stages of development. Ductal plates are layers of cells that form the basis for the development of bile ducts. You may hear this called 'ductal plate malformation'.

The signs, symptoms and complications for each condition can vary widely. They almost always involve the development of cysts or unusually shaped bile ducts.

Due to the different organs involved, your child may see a range of professionals. This may include:

- liver doctors (hepatologists)
- kidney doctors (nephrologists)
- specialist nurses
- radiologists
- surgeons

What are cysts?

Cysts are pockets of tissue filled with fluid that develop inside an organ. They can grow larger over time. This may reduce the ability of an organ to carry out its normal job. It can also cause a range of symptoms.

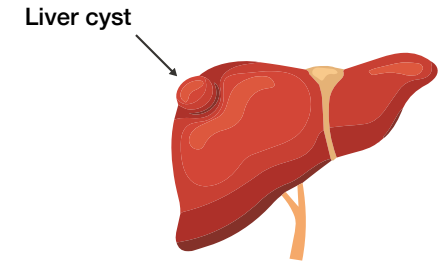


Figure 2: Liver cyst

Congenital hepatic fibrosis

What is congenital hepatic fibrosis?

Congenital hepatic fibrosis (CHF) is a rare genetic condition. It happens in around 1 in every 20,000 births.

If CHF happens on its own, it is called isolated congenital hepatic fibrosis. This is very rare. In most cases, CHF happens alongside other diseases that affect the liver and kidneys. This is why liver doctors, when making a diagnosis of CHF in a child, will always screen for kidney disease.

CHF is generally not associated with cysts. The main problem in this condition is stiffening of the liver. This is caused by liver scarring (fibrosis). This scarring generally gets worse as children and young people get older. But the speed and severity of scarring will vary from child to child. Most cases first show in children and teenagers. But some people are diagnosed as adults.

In CHF, scarring mainly affects the structure of the liver and not the liver cells. This means that the liver will usually keep doing its job. This is why some children show no symptoms.

In children with congenital hepatic fibrosis, scarring (fibrosis) of the liver can become worse over time. This can lead to severe scarring, known as cirrhosis. This may happen in adulthood.

In some children, when CHF is diagnosed, scans also reveal cysts or unusually shaped bile ducts. This is called Caroli syndrome (see page 11).

How is congenital hepatic fibrosis diagnosed?

CHF may only be diagnosed when your child is being tested for something else. A diagnosis may also happen if they have portal hypertension (see page 10). This is a complication of liver scarring.

Tests used to help with the diagnosis may include:

- physical examination
- blood tests
- genetic tests
- abdominal ultrasound scan
- liver biopsy
- endoscopy (if portal hypertension is suspected)
- magnetic resonance imaging (MRI) / magnetic resonance cholangiopancreatography (MRCP)
- endoscopic retrograde cholangiopancreatography (ERCP)

What are the symptoms of congenital hepatic fibrosis?

Children can start getting symptoms at any age. Some children never have symptoms.

Symptoms are often caused by a complication called portal hypertension. Symptoms may also be caused by infections in the bile ducts. Even if your child has symptoms, their liver will usually still be working well.

Symptoms may include:

- enlarged liver (hepatomegaly)
- enlarged spleen (splenomegaly)
- tummy (abdominal) pain
- vomiting blood (haematemesis)
- black, “tarry” poo (stools)
- high temperature (fever)
- yellowing of whites of the eyes and skin (jaundice)
- being sick (vomiting)
- gallstones (cholelithiasis)



What are the possible complications of congenital hepatic fibrosis?

Possible complications include:

Liver scarring (fibrosis)

In children with CHF, scarring (fibrosis) of the liver can become worse over time. This can lead to severe scarring, known as cirrhosis. The speed and severity of scarring varies from child to child. Your child may never develop cirrhosis or they may develop it as an adult.

When scarring happens inside the liver, it stiffens. This makes it more difficult for blood to flow through it. This can cause high pressure in the portal vein.

Portal hypertension

High pressure in the portal vein is also known as portal hypertension. Portal hypertension causes swollen blood vessels in the food pipe, stomach and intestine. These blood vessels have thin walls and may bleed. This can cause your child to vomit blood or pass black tarry poo (stools). Portal hypertension can also cause fluid to build up in the tummy (abdomen). This is known as ascites.

Cholangitis

This is inflammation (swelling) and infection of the bile ducts. This mainly occurs when CHF is linked with cysts or problems with the bile ducts. This is known as Caroli syndrome (see page 11).

How is congenital hepatic fibrosis treated?

There is no cure or specific treatment for congenital hepatic fibrosis. Medical care focuses on reducing symptoms and managing complications. Treatment varies from child to child depending on the severity of symptoms.

Treatments for portal hypertension and its complications may include:

- endotherapy to treat swollen blood vessels in the food pipe
- medication to help reduce blood pressure in the portal vein
- transjugular intrahepatic portosystemic shunt (TIPS)
- liver transplant

Treatments for bile duct infection (cholangitis) and its complications may include:

- antibiotics injected into a vein (intravenous antibiotics)
- draining fluid from cysts (cyst aspiration) to reduce their size
- endoscopic retrograde cholangiopancreatography (ERCP) to treat narrow or blocked bile ducts
- percutaneous transhepatic cholangiogram (PTC) to treat narrow or blocked bile ducts
- liver transplant when there is repeated bile duct infection (cholangitis) and/or severe sepsis

Caroli disease and Caroli syndrome

What are Caroli disease and Caroli syndrome?

Caroli disease and Caroli syndrome are both rare, genetic conditions. They affect the larger bile ducts that can be found inside and outside the liver. The two conditions are often grouped together because they share some features. But there are also some differences.

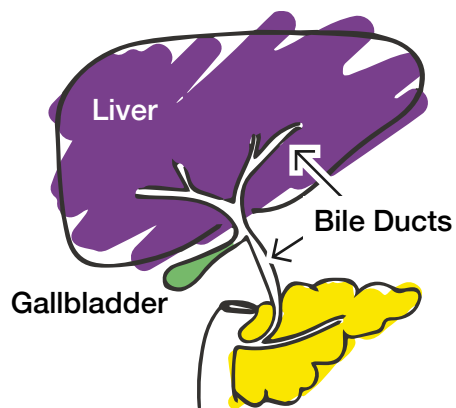
Caroli disease

Caroli disease happens in around 1 in every 1 million births. It usually happens at random, but it has been known to run in families.

The main features of Caroli disease are:

- cysts inside the larger bile ducts within the liver
- widening (dilation) of the larger bile ducts inside the liver

Figure 3:
Location of the
bile ducts



If the bile ducts become too wide, bile can collect and cause swelling and infection. This is known as cholangitis. The swelling stops bile from getting out of the liver. If the bile builds up, it can form stones. Often, these stones won't cause problems. In a small number of people, they can block the bile duct and cause symptoms.

Caroli syndrome

Caroli syndrome is more common than Caroli disease. It happens in around 1 in every 100,000 births.

Caroli syndrome and Caroli disease share a key feature. In both conditions, children are born with cysts and widening of the bile ducts inside the liver.

But Caroli syndrome has some differences. This condition also features congenital hepatic fibrosis (see page 7). This means that children with Caroli syndrome have scarring and stiffening of the liver. They also have the symptoms and complications associated with CHF. This includes portal hypertension (see page 10).

Children and young people with Caroli syndrome may also develop cysts on their kidneys. This is why Caroli syndrome is often linked with autosomal recessive polycystic kidney disease (ARPKD). In this genetic condition, multiple cysts develop in the kidneys.

How are Caroli disease and Caroli syndrome diagnosed?

Tests used to help with the diagnosis may include:

- physical examination
- blood tests
- genetic tests
- abdominal ultrasound scan
- magnetic resonance imaging (MRI) / magnetic resonance cholangiopancreatography (MRCP)
- endoscopy (if portal hypertension is suspected)



What are the symptoms of Caroli disease and Caroli syndrome?:

The symptoms of Caroli disease and Caroli syndrome can appear at any age. But symptoms are most likely to appear in teenagers and young adults.

Symptoms are usually linked to bile duct infection (cholangitis) and the development of gallstones (cholelithiasis).

Symptoms may include:

- high temperature (fever)
- tummy (abdominal) pain
- yellowing of the whites of the eyes and skin (jaundice)
- being sick (vomiting)



If your child has Caroli syndrome, there may also be signs of portal hypertension. Extra symptoms may include:

- enlarged spleen (splenomegaly)
- tummy (abdominal) pain
- vomiting blood (haematemesis)
- black, “tarry” poo (stools)

What are the possible complications of Caroli disease and Caroli syndrome?

Possible complications include:

- repeated infection of the bile ducts (cholangitis)
- narrowing of the bile ducts (strictures)
- liver abscesses
- infection of the bloodstream (septicaemia)
- stones in the bile ducts (cholelithiasis)
- liver failure
- slightly increased risk of developing bile duct cancer (cholangiocarcinoma)

Extra complications in Caroli syndrome may include:

- portal hypertension
- ascites

How are Caroli disease and Caroli syndrome treated?

There are no cures or specific treatments for Caroli disease and Caroli syndrome. Medical care focuses on reducing symptoms and managing complications. Treatment varies from child to child depending on the severity of symptoms.

Treatments for portal hypertension and its complications may include:

- endotherapy to treat swollen blood vessels in the food pipe
- medication to help reduce blood pressure in the portal vein
- transjugular intrahepatic portosystemic shunt (TIPS)
- liver transplant

Treatments for bile duct infection (cholangitis) and its complications may include:

- antibiotics injected into a vein (intravenous antibiotics)
- draining fluid from cysts (cyst aspiration)
- endoscopic retrograde cholangiopancreatography (ERCP) to treat narrow or blocked bile ducts
- percutaneous transhepatic cholangiogram (PTC) to treat narrow or blocked bile ducts
- liver transplant when there is repeated bile duct infection (cholangitis) or severe sepsis

Biliary hamartomas

What are biliary hamartomas?

Biliary hamartomas are very small, dense cysts. They are usually scattered all over the liver. They are rare and affect less than 1 in 100 children. They are not a type of cancer and do not cause any problems.

Biliary hamartomas are also known as ‘bile duct hamartomas’ or ‘Von Meyenburg complexes’. They are often found in normal livers. Most children have no symptoms. The liver will continue to work, and no treatment or monitoring is needed.

Biliary hamartomas can happen alongside other diseases that affect the liver and kidneys. They are more common in children and young people with:

- autosomal dominant polycystic kidney disease (ADPKD)
- congenital hepatic fibrosis (CHF)
- Caroli syndrome

How are biliary hamartomas diagnosed?

Biliary hamartomas are often found by accident. This can happen when doctors are doing scans to look for something else. The cysts may be mistaken for cancer when they are first found. Your child will have more tests to confirm the diagnosis.

Tests used may include:

- abdominal ultrasound scan
- magnetic resonance imaging (MRI) or magnetic resonance cholangiopancreatography (MRCP)



What are the symptoms of biliary hamartomas?

Most children show no signs or symptoms. If symptoms do happen, they may be similar to the symptoms for bile duct infection (cholangitis). They can include:

- high temperature (fever)
- tummy (abdominal) pain
- yellowing of the whites of the eyes and skin (jaundice)
- being sick (vomiting)

How are biliary hamartomas treated?

Biliary hamartomas do not usually cause problems. In most children, the liver continues to work normally, and no treatment is needed. Treatments will only be used if your child develops symptoms that cause concern.

What are the possible complications of biliary hamartoma?

Biliary hamartomas are not a type of cancer. But there is a very small risk they could transform and progress to bile duct cancer (cholangiocarcinoma).

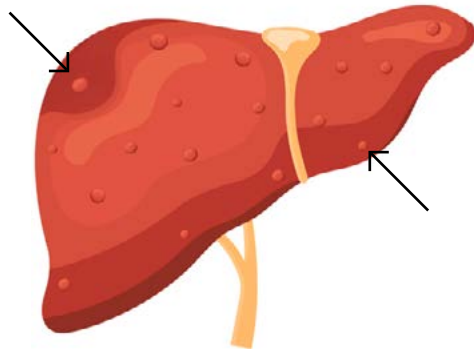
Polycystic liver disease

What is polycystic liver disease?

Polycystic liver disease (PLD) is a condition in which 10 or more cysts grow inside the liver. The cysts are not a type of cancer. In children and young people, the cysts are usually very small.

In a small number of people the cysts grow and increase in number with age. This is why symptoms may only appear in adulthood.

Figure 4:
Liver cysts



Polycystic liver disease can run in the family (be inherited). But people can still have different experiences of it. Some people may become very unwell, but others will have no problems, even though they are related.

There are two forms of polycystic liver disease:

Isolated polycystic liver disease

Polycystic liver disease can happen on its own without kidney disease. This is sometimes called autosomal dominant polycystic liver disease (ADPLD). In this type of PLD, cysts only happen in the liver. This type is very rare.

Polycystic liver disease with polycystic kidney disease (PKD)

Polycystic liver disease usually happens as part of a condition called autosomal dominant polycystic kidney disease (ADPKD). In this type, cysts grow in the liver and in the kidneys. Around 80 or 90 out of every 100 people with ADPKD will also have polycystic liver disease.

How is polycystic liver disease diagnosed?

Polycystic liver disease is often found by accident when doctors are doing scans to look for something else.

Tests used to help with the diagnosis may include:

- abdominal ultrasound scan
- magnetic resonance imaging (MRI) scan
- blood tests
- genetic tests

What are the symptoms of polycystic liver disease?

Most children and young people show no signs or symptoms. Even if your child has a lot of cysts in their liver, it will carry on working as normal. The liver cells around the cysts are not damaged.

Even in adulthood, most people have no symptoms from liver cysts. Symptoms only happen if the liver cysts grow larger over time.

What are the possible complications of polycystic liver disease?

It is very unusual for complications to happen in children and young people. But in a small number, the liver cysts get larger as they age. This happens in around 1 in 20 people with PLD. If the number, or size of cysts gets very big, it can cause symptoms. This is due to the space the cysts take up in the tummy (abdomen). Symptoms may include:

- tummy (abdominal) pain
- tummy (abdominal) swelling
- back pain
- feeling short of breath (dyspnoea)
- feeling full early in a meal
- indigestion (dyspepsia)
- heartburn (gastro-oesophageal reflux)

In rare cases, the cysts get so big that the liver grows and pushes on other organs and blood vessels. This can lead to further complications including:

- enlarged liver (hepatomegaly)
- portal hypertension and ascites

- cyst infection
- cyst bleeding
- cyst rupture (bursting)
- yellowing of the whites of the eyes and skin (jaundice)

How is polycystic liver disease treated?

Polycystic liver disease does not usually cause problems in children and young people. It is very rare for them to need treatment. This will only be needed if your child develops serious symptoms that cause concern.

Treatments in rare cases may include:

- antibiotics for cyst infection
- draining fluid from cysts (cyst aspiration)
- medication to slow down cyst growth
- surgery – procedures such as removing the wall from cysts (cyst fenestration), liver resection and liver transplant.

If your child is diagnosed with polycystic liver disease, they will be monitored over time. This is done to check for any changes in the size or number of cysts.

As they age, hormones may play a role in the number and size of cysts. In adulthood, women are more likely to have symptoms and need treatment. The medical team may recommend avoiding contraceptives that contain oestrogen. This is because oestrogen can increase the growth of liver cysts.

Useful resources

Glossary of liver terms

Many medical terms may be used by those caring for children and young people with liver disease. Follow this QR code for a list of terms.



Other CLDF resources

CLDF produces a wide range of information for children and young people up to the age of 25 with liver disease, their families and the healthcare professionals who look after them.

Information can be downloaded or ordered from:
 Website: www.childliverdisease.org
 Email: info@childliverdisease.org
 Tel: 0121 212 3839
 Follow this QR code to see our resources.



Information from other organisations

Polycystic Kidney Disease Charity

www.pkdcharity.org.uk

A UK charity dedicated to improving the lives of individuals and families affected by polycystic kidney disease (PKD). They provide information, advice and personalised support. They also raise awareness and fund research.

How we produce information

Our information resources are written, edited and reviewed with the help of medical experts and families living with liver disease.

Thanks

We would like to thank staff at each of the specialist paediatric liver centres: Birmingham Children's Hospital, King's College Hospital and Leeds Children's Hospital. We would also like to thank the families who reviewed our information.

PIF TICK accreditation

We have gained PIF TICK accreditation for our information production process. Follow this QR code for more details.



Disclaimer

This resource provides general information but does not replace medical advice. It is important to contact the medical team if you have any worries or concerns.

Feedback and Information Sources

Your feedback is important to us and will help us improve our information. To provide feedback, or for more information on how it was developed, email info@childliverdisease.org.

This leaflet was reviewed in February 2025

It is due to be reviewed by February 2028

Children's Liver Disease Foundation

Children's Liver Disease Foundation (CLDF) is the UK's leading organisation dedicated to fighting all childhood liver diseases.

CLDF provides information and support services to young people up to the age of 25 with liver conditions and their families and is a voice for everyone affected.

CLDF is here for you, whether you want to talk, share with others, or just belong to a group which cares, knows what it's like and is fighting to make a difference.

Are you a young person up to the age of 25?

Contact the Young People's Team:

Phone: 0121 212 6024 **Email:** youngpeople@childliverdisease.org

Are you the parent or carer of a child or young person with a liver condition?

Contact the Families Team:

Phone: 0121 212 6023 **Email:** families@childliverdisease.org

Would you like to help us support the fight against childhood liver disease?

Please help us to continue to support children, young people and families now and in the future.



Contact the Fundraising Team:

Email: fundraising@childliverdisease.org

Use this QR code to donate

Tel: 0121 212 3839

Email: info@childliverdisease.org

Website: www.childliverdisease.org

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