Children's Liver Disease Foundation

fighting childhood liver disease

Primary Sclerosing Cholangitis

A Guide

An explanation of what primary sclerosing cholangitis is, its causes, diagnosis and treatment

This information has primarily been written for:

- Parents/carers of a child diagnosed with primary sclerosing cholangitis (PSC)

Others who may also find this information useful:

- Young people diagnosed with PSC
- Healthcare professionals who would like to find out more about the condition.

This leaflet aims to:

- Explain what PSC is
- Discuss the signs and symptoms
- Discuss the diagnosis, treatment and possible complications.

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

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

What is primary sclerosing cholangitis?

Primary sclerosing cholangitis (PSC) is a rare, progressive liver disease in which bile ducts inside (intrahepatic) and outside (extrahepatic) the liver may narrow or become blocked due to inflammation and scarring.

In a healthy, functioning liver, bile is made by liver cells and then transported through the bile ducts to the gallbladder. When a meal is eaten, the gallbladder contracts and bile flows into the intestine. This allows toxins to be eliminated from the body in the stool. Bile in the intestine also helps with the absorption of some fats and essential vitamins (vitamins A, D, E and K). In PSC, the bile ducts become progressively scarred and narrow so that bile is unable to flow normally. This allows toxins to accumulate and damage the biliary tree and the liver.

PSC is a rare condition with a prevalence of 1.5 cases per 100,000 children. PSC may occur alone but is more common in children with inflammatory bowel disease (IBD), affecting at least 10% of those with ulcerative colitis.

What causes PSC?

The cause of PSC is unknown but researchers suggest there are both genetic and environmental factors involved.
Current research suggests that PSC may be associated with an immune reaction against the liver generated by changes in the gut (an autoimmune condition). This leads to inflammation causing scarring and subsequently damage to the bile ducts.
Other research suggests that there may be an imbalance in the microorganisms or bacteria (gut flora) in the bowel.

What are the signs and symptoms of PSC?

A child may have PSC for years before they develop symptoms. In some children, it may be identified due to routine blood tests, medical investigations for IBD or vague symptoms such as tiredness.

As with most diseases, there is a range of severity of symptoms. Most children and young people have no symptoms if the disease is mild, whereas in advanced disease there may be:

- Jaundice (yellowing of the skin and eyes)
- Itching
- Dark coloured urine and pale stools (usually occurs if the bile ducts are blocked)
- Infection of the bile duct (cholangitis)
- Tiredness and reduced energy
- Loss of appetite
- Weight loss
- Abdominal pain
- A high temperature (in rare cases).

How is PSC diagnosed?

If a child or young person is displaying symptoms of PSC, there are number of tests which may be used to confirm a diagnosis:

Family history

There may be other people in the family with other autoimmune diseases (such as diabetes, thyroid disease, coeliac disease or IBD).

Blood tests

Liver blood tests may be carried out over multiple days or weeks to show a pattern that would be in keeping with the bile ducts being damaged (ALP, GGT or bilirubin being raised). These blood results are suggestive but not diagnostic.

Blood tests for autoantibodies may show a positive p-ANCA result. Autoantibodies can also be found in some other autoimmune diseases. Again, this is not diagnostic.

Liver blood tests may also indicate if the liver is not working normally due to the damage caused. This may include clotting tests such as INR and measuring albumin levels.

Ultrasound scan

An ultrasound is often a key diagnostic tool in liver disease. It may show an enlarged gallbladder, areas of dilated bile ducts or areas of narrowing.

An ultrasound can also show changes associated with advanced liver disease such as fluid in the abdomen (ascites) and an enlarged spleen (portal hypertension).

Magnetic resonant cholangio-pancreatogram (MRCP scan)

This is a specialist magnetic resonance imaging (MRI) scan of the bile ducts. It can show the changes in the bile ducts inside the liver by using a special contrast dye to identify these areas on the scan. This is better at diagnosing PSC than an ultrasound.

Liver biopsy

A liver biopsy may be useful if the diagnosis is not clear from the above tests. A biopsy can show scarring around the bile ducts and bile duct damage. The biopsy may also show how much scarring there is in the liver (fibrosis). Advanced fibrosis is known as cirrhosis.

Further information about the signs of liver disease, a glossary of terms and routine investigations can be found in the CLDF leaflet "An Introduction to Liver Disease".

Can PSC be treated?

There is currently no definitive treatment for PSC. Supportive treatment is focused on managing symptoms and improving bile flow and the consistency of bile in the bile ducts.

One of these symptoms may be pruritus. This is an intense itch beneath the skin that can cause severe discomfort and can be difficult to control. The cause is not completely understood but is thought to be due to increased levels of bile acids/salts in the blood. Management of pruritus will be discussed by your multi-disciplinary medical team and treatment given accordingly.

In children where PSC is associated with IBD, improvement of the bowel disease may also improve the liver.

Medicines

- **Ursodeoxycholic acid (UDCA)**
  This can improve bile flow out of the liver which may reduce jaundice and/or itching. Data from adult patients suggests that UDCA does not change the natural history of the disease.

- **Antibiotics**
  If an infection has occurred in the bile ducts (cholangitis), antibiotics are used to reduce the risk of the infection occurring again.

Medication and treatment options may vary and your child's medical team will discuss the most appropriate options available.

As with other chronic liver diseases, medicines such as aspirin and ibuprofen should be avoided as they can make bleeding in the gut more likely. Paracetamol (Calpol) is a safer alternative.

Vitamins

Additional vitamin supplements may be necessary because poor bile flow can reduce the absorption of fat-soluble vitamins i.e., A, D, E and K. These are normally given orally, although an injection may be needed.

Diet

The development of fluid in the abdominal cavity (ascites) can occur, but usually only in patients with advanced disease (cirrhosis). When ascites occurs, a low salt diet is sometimes recommended but management will most likely involve the use of diuretics to increase excretion of the excess fluid through the urine. Advice and support will be provided by your dietetic team if changes to your child's diet are required.

Endoscopic procedures

In a small number of cases endoscopic procedures may be needed. These include:

- Children and young people with IBD who will need regular colonoscopies.
- Children and young people with cirrhosis who may need a gastroscopy to look for dilated blood vessels in the gullet (oesophagus) or stomach. These dilated vessels (varices) are a result of a condition called portal hypertension.
- An endoscopic retrograde cholangio-pancreatography (ERCP) may be required to make the diagnosis in certain individuals if their MRI scan is inconclusive. This is usually performed under general anaesthetic at a specialist centre.

  An ERCP is more often used to treat blockages (strictures) in the bile duct and can involve the insertion of a plastic or metal tube (stent). Brushings of strictures are usually taken to allow the cells to be assessed. After the insertion of a stent, the flow of bile will be improved. ERCPs are rarely required in children but takes place more often in adolescents. Your specialist centre will assess the need for this procedure.

What happens if the disease progresses?

If the disease progresses, the following complications may be experienced:

- Biliary strictures - narrowing of a bile duct from scar tissue within the duct itself. A stricture can block the release of bile and lead to infection (cholangitis). An ERCP may be required.
- Progressive itch
- Portal hypertension – high blood pressure in the portal vein which may occur due to scarring of the liver or a blockage in the portal vein. The spleen may become enlarged (splenomegaly) due to a back flow of blood from the liver and cause abdominal distension.
- Varices - swollen veins in the intestine, oesophagus and stomach which can bleed if not treated. An endoscopy is usually required.
- Ascites – collecting of fluid in the abdomen. Ascites can be managed by diuretic medicines to increase urine output and minimise fluid accumulation.

  Further information about portal hypertension, varices and ascites is available in other CLDF leaflets.

- Weight loss
- Development of bile duct cancer (cholangiocarcinoma). This is extremely rare in children.

The role of liver transplantation

A liver transplant may be discussed as a treatment option if the liver begins to fail or if there is severe recurrent cholangitis. If a transplant is the best treatment option, the medical team will focus on preventing complications and treating symptoms while waiting for an organ.

Outcomes after transplantation for PSC are positive and many children go on to have a good quality of life. However, there is a chance that PSC can reoccur in the transplanted liver. The underlying reasons for this are not yet fully understood. Risk factors taken from studies of adult patients include a younger age at transplant and the co-existence of IBD.

Further information is available in the CLDF leaflet "Liver Transplantation".

Are there any restrictions on lifestyle?

Many children with PSC can live a relatively normal quality of life.

Severe itching may disturb sleep and contribute to fatigue, tiredness and irritability. Reduced appetite, nausea and vomiting may also impact their daily lives.

There is a CLDF leaflet called "Pruritus" that has further information and tips from parents on how to manage this.

If and when fatigue is experienced, this can impact the level of physical activity your child is able to carry out on a day-to-day basis and possibly their school life. It is best to talk to your child's school about their needs in these circumstances.

If your child has an enlarged spleen, it is advised that contact sports should be avoided due to the risk of bleeding from a splenic injury. Please talk to your medical team about this as it depends on the individual.

CLDF have an Education Pack to help families discuss the needs of their child with schools. Contact our Family Support Team for further information at **families@childliverdisease.org** or by calling **0121 212 6028**.

Living with a chronic illness and dealing with the side effects of treatment can be distressing. Specialist psychology teams at liver centres may be able to provide help and support. The CLDF Support Team can also signpost and refer you to the most appropriate services.

What happens in the future?

Your child's growth and development will be followed up by periodic visits to hospital. How often you will need to go to hospital varies for each child. These visits may occur more frequently if your child's disease appears to be active (e.g. persistently abnormal blood results or other tests) and/or progressing.

Children's Liver Disease Foundation (CLDF) is in touch with families who have a child with PSC. PSC does not have to hold your child back from reaching their full potential. You can contact CLDF for further support and details can be found on the back of this leaflet.

You may benefit from contacting PSC Support (pscsupport.org.uk). They are a UK based charity and although they work more widely with adults, they are a good source of safe information and have useful videos and events specifically for patients with PSC.

Is research taking place?

The slow progressive nature of PSC has made clinical trials more difficult but new treatments are constantly under investigation.

There are multi-centre clinical trials for paediatric patients taking place internationally to find answers. Furthermore, there are several areas of active investigation in this field with promising clinical data from adults with PSC. Personalised medicine and treatments tailored to individual patients may be the future of PSC therapy.

Notes

CLDF produces a wide variety of information resources for children and young people up to the age of 25 with liver disease, their families and the healthcare professionals who look after them. This information can be downloaded or ordered from CLDF's website childliverdisease.org. For further enquiries regarding CLDF's information please contact the Information and Research Hub Manager by email at irhm@childliverdisease.org or call 0121 212 6029.

Thanks

This booklet has been written, edited and reviewed with the help of staff at each of the specialist paediatric liver centres: Birmingham Women's & Children's Hospital, King's College Hospital and Leeds Children's Hospital. Thank you to all the staff involved who have made the production of this leaflet possible.

Disclaimer

This leaflet provides general information but does not replace medical advice. It is important to contact your/your child's medical team if you have any worries or concerns.

Feedback and Information Sources

Information within this leaflet has been produced with input from the three specialist paediatric liver centres in the UK. To provide feedback on this leaflet, or for more information on the content of this leaflet including references and how it was developed contact Children's Liver Disease Foundation: info@childliverdisease.org.

This leaflet has been developed in June 2021. It is due to be reviewed by June 2024.

What is Children's Liver Disease Foundation (CLDF)?

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, funds vital research into childhood liver disease and is a voice for everyone affected.

Are you a young person up to the age of 25 with a liver condition or a family member? CLDF's Families and Young People's teams are here for you, whether you want to talk about issues affecting you, meet and share with others, or just belong to a group which cares, knows what it's like and is fighting to make a difference. You are not alone.

If you are a parent/carer or family member then get in touch with CLDF's Families Team:
Phone: 0121 212 6023 **Email:** families@childliverdisease.org

If you are a young person and want to find out more about CLDF's services you can contact CLDF's Young People's Team:
Phone: 0121 212 6024 **Email:** youngpeople@childliverdisease.org

CLDF have a social media platform called HIVE/HIVE+ for 13-24 year olds with a liver disease/transplant to make new friends, connect and share stories. **childliverdisease.org/young-people/hive**

Would you like to help us support the fight against childhood liver disease? CLDF's work relies on voluntary donations and fundraising. Please help us to continue to support children, young people and families now and in the future. To find out more about fundraising and how you can join the fight against childhood liver disease you can visit childliverdisease.org/support-us. Alternatively, you can email the Fundraising Team at fundraising@childliverdisease.org or call them on **0121 212 6022.**

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