Polycystic Liver Disease (PLD)

What is PLD?

Polycystic Liver Disease (PLD) is a genetic disease, which means that you probably inherited it from one of your parents. This means that other people in your family could also have PLD. Genetic diseases are caused by mutations (small changes) in your DNA. In PLD, these mutations cause liver cysts to develop. Patients with PLD have multiple (>10) cysts in their liver; a cyst is a closed sac filled with fluid. The buildup of multiple cysts and the growth of single cysts may cause the liver to grow. Many patients with PLD also have cysts in their kidneys.

Who is affected?

Many patients with PLD don't have any symptoms at all. Their cysts are diagnosed by coincidence. Of the PLD patients with symptom, approximately 80% are women and PLD is usually diagnosed between ages 30 to 40.

Why do my cysts grow?

Within patients with PLD, there are big differences in the rate of cyst growth. The only known factor that increases cyst growth rate is estrogen, the female growth hormone. This is the reason why most patients with symptoms are female and why the fastest cyst growth is seen in fertile women. Growth rate slows down when estrogen levels drop after the menopause.

Which symptoms can occur?

The majority of patients do not have any complaints from liver cysts. However, persistent growth of the cysts may lead to enlargement of the liver, up to 10 times its normal size.
Liver enlargement may cause symptoms such as abdominal fullness, lack of appetite, early satiety, acid reflux, nausea, pain in the liver area and deferred pain such as in the shoulder, shortness of breath, limitations in mobility, fatigue, anxiety and discontentment with appearance, particularly due to distension of the stomach.

How is PLD diagnosed?

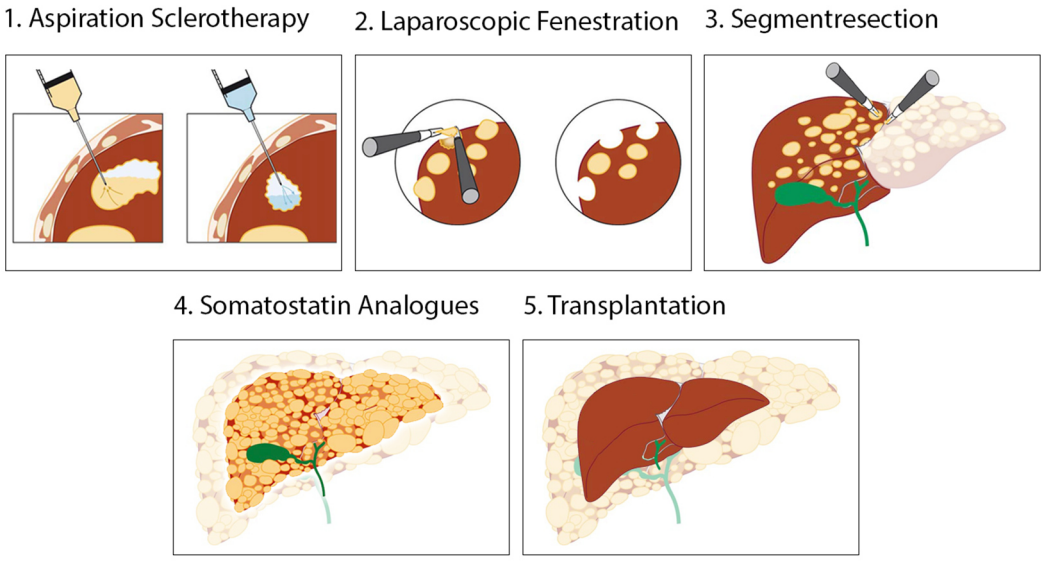
Your doctor can make a diagnosis with an ultrasound, CT-scan or MRI. When there are 10 or more cysts in the liver, the diagnosis of polycystic liver disease is made.

What can my doctor do?

As hepatic cysts normally do not cause any trouble, it is often not necessary to treat them.

However, several treatment options are available in the event of symptoms:

1) Treatment options include puncturing large cysts and 'burning' the cyst wall with for example ethanol or polidocanol. This procedure is often called Aspiration Sclerotherapy.
2) Cysts can also be removed with two types of surgery. In a procedure called cyst fenestration, a surgeon drains and removes cysts with key-hole surgery.
3) In the other option, called segmentresection, a very specialized surgeon removes a part of the liver.
4) Medication with somatostatin analogue injections can be used to slow cyst growth if there are numerous small cysts and an enlarged liver.
5) The last resort in a small proportion of severely affected patients is liver transplantation.



As doctors are not necessarily familiar with PLD, they may not have much information to give you. Therefore, you can ask your doctor to discuss your case with experts of the ERN if there are no PLD-experts available in your region or country.

If you also have kidney cysts, you should see a nephrologist or kidney specialist.

What can I do?

Medication:

We advise patients to not use any medication with estrogen, since female hormones in birth control and postmenopausal hormone therapy lead to faster liver growth.

Diet:

There is no special diet for PLD. We recommend that you eat a healthy, well balanced diet. If you also have kidney cysts, we recommend a low salt diet (< 6 grams/day) and a fluid intake of about 3 liters/day. This can help protect the kidneys.

Family planning:

You can consult your doctor for advice on family planning and contraception. Your doctor may refer you for genetic consultation.

Will my liver function be affected?

The liver is an organ with many different functions. Even with many cysts present in the liver, it will still remain fully functional. Liver-related blood tests (i.e. GGT and ALP) can be higher than normal values in patients with PLD. Importantly, this is not a matter of concern.

What are the complications of PLD?

Complications of PLD include infection, bleeding or rupture of cysts. When cysts are located near one of the important liver veins, this vein might be compressed. This can cause the accumulation of fluid in the belly.

Even though complications of cysts are rare, these are all serious complications that require urgent treatment. Therefore, we advise you to consult your physician if you experience any of the following symptoms: fever, acute onset severe liver pain or unexplained rapid increase in belly size.

Do I need medical checkups?

PLD patients without any symptoms do not require any standard medical checkups. In patients with symptoms that undergo treatment, ultrasound, CT-scans or MRI-scans are used for follow-up.

Do my family and I need to get genetically tested?

Outcomes of genetic testing will not influence the treatment that you will receive, but genetic testing might help you or your children in choosing appropriate contraception. However, please keep in mind that finding a genetic cause for PLD might increase your premiums for a life insurance or mortgage. Since not all genes that cause PLD are discovered yet, genetic testing does not always find the mutation that causes PLD.

Will I develop cancer in my cysts?

No, PLD is a benign disease and the cyst cells will not turn into cancer. Cysts may however, grow bigger throughout your life which may cause symptoms.

How can I find a specialist?

PLD may need to be managed by a doctor or a hospital network with experience or an interest in PLD. PLD is a rare disease and not every doctor has an interest in or experience of PLD. However, some hospitals are part of the Europe-wide network for rare liver diseases, ERN RARE-LIVER. This means that patients seen at hospitals within the network can benefit from the expertise of specialists who work at other hospitals within the network. For more information about the ERN RARE-LIVER, visit https://rare-liver.eu/.

Find information and patient support

Click on the section "patients" on the ERN RARE-LIVER website (https://rare-liver.eu/).

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