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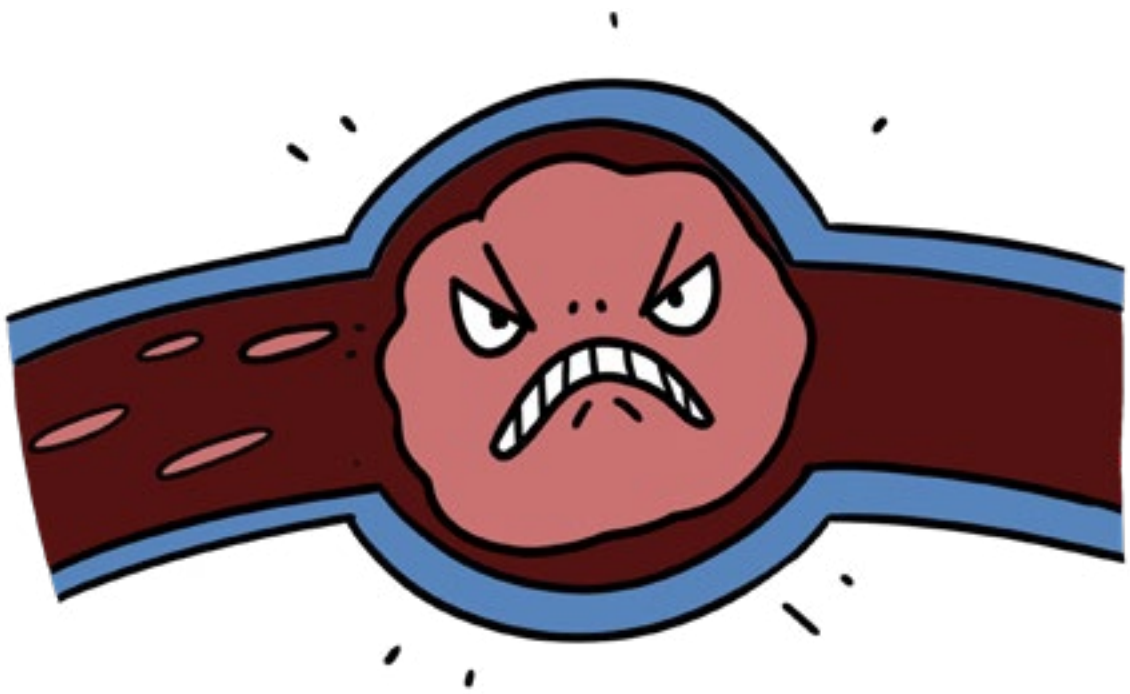
European Reference Network on Hepatological Diseases

BUDD CHIARI SYNDROME (BCS)

What is BCS?

Budd-Chiari syndrome is a condition in which the hepatic veins (veins that drain the liver) are blocked or narrowed by a clot (mass of blood cells).



Representation of a Clot in a vessel

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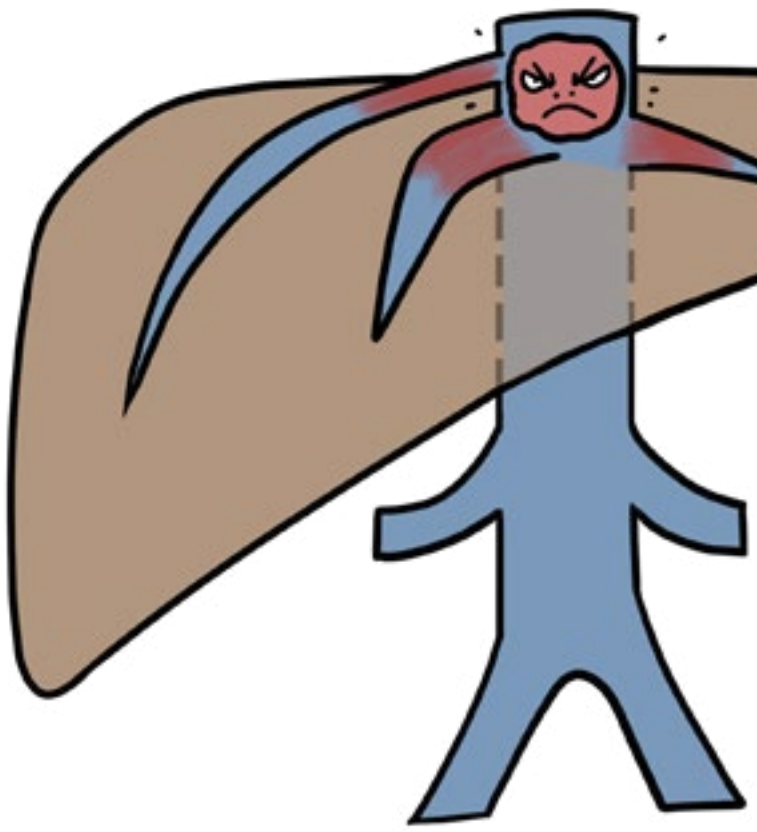

Clot in hepatic veins or inferior vena cava=Budd chiari syndrome

What are the symptoms?

This blockage causes blood to back up into the liver, and as a result, the liver grows larger. The spleen (an organ located on the upper left side of the abdomen that helps fight infection by filtering the blood) may also grow larger.

Symptoms are varied and can be:

- No symptoms
- Fatigue
- Upper abdominal pain (liver area)
- Accumulation of fluids in the abdomen (ascites)
- Leg oedema/swelling
- Oesophageal varices diagnosed on endoscopy
- less frequent symptoms can be:

  Gastrointestinal bleeding from oesophageal varices
  Jaundice (skin, whites of the eyes and mucous membranes turn yellow)
  Acute worsening of liver function

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Ascites

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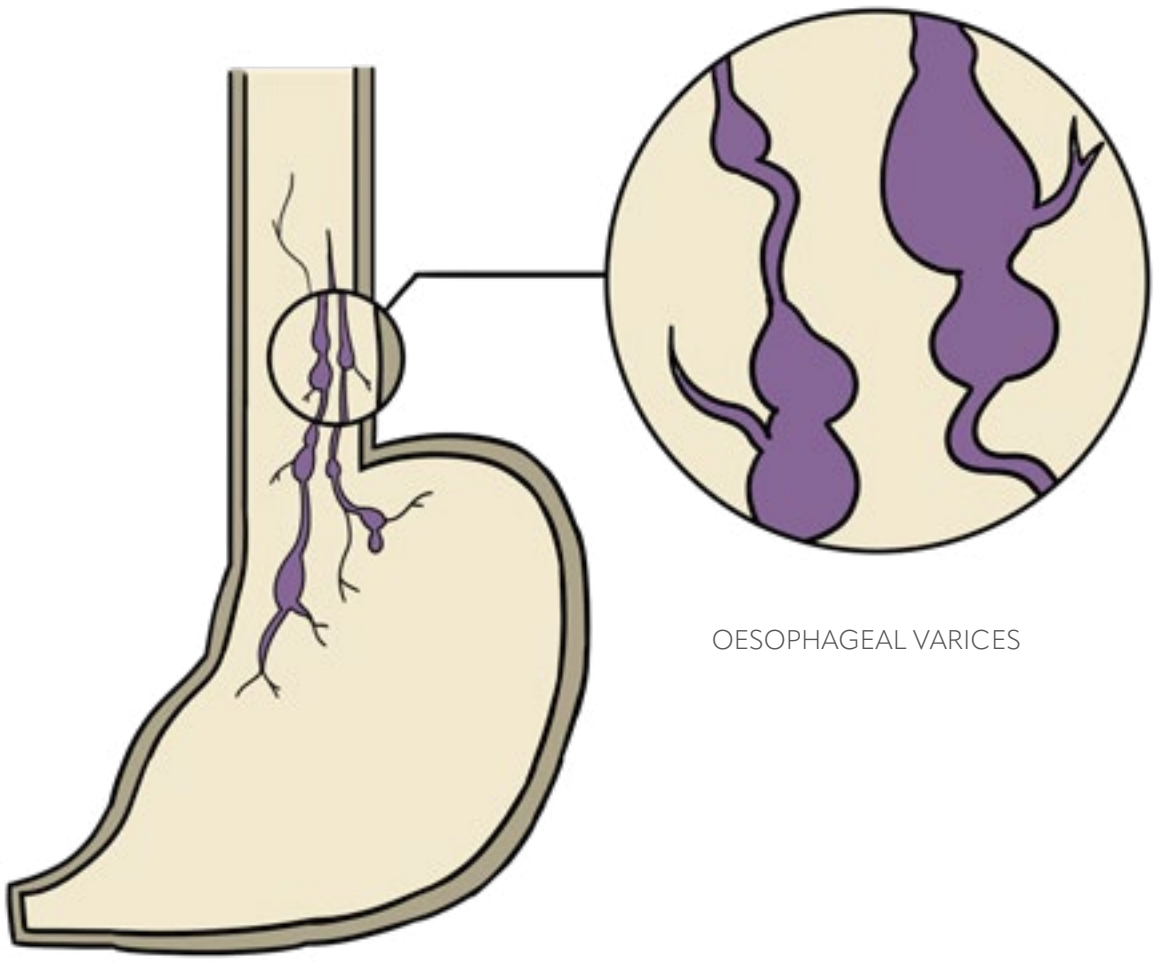

OESOPHAGEAL VARICES

How is BCS diagnosed?

Imaging tests will determine if your veins are blocked by clots. These tests include:

- Ultrasound with Doppler study: a procedure that transmits high-frequency sound waves through body tissues. The echoes are recorded and transformed into video or photographs of the internal structures of the body. Doppler study allows to see the vessels that goes to and move away from the liver. If a clot exists, usually it is seen with this technique.
- Computed tomography (CT) scan: uses X-rays and computers to produce images of a cross-section of the body.
- *Magnetic resonance imaging (MRI):* test that uses a large magnet, radio waves, and a computer to produce very clear pictures of the body.

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What are the causes?

There is often a combination of several predisposing **risk factors**. The main one is the blood disorder, myeloproliferative disorder (overproduction of white blood cells, red blood-cells or platelets). **Myeloproliferative disorder** is an acquired disease; it is not transmitted by parents at birth.

Other predisposing risk factors are acquired or **congenital anomalies**. The main congenital anomalies are the natural anticoagulant deficiency (protein C, protein S, antithrombin) and mutation of factor V Leiden or prothrombin gene mutation.

There are other disorders that shall always be searched for, particularly autoimmune, as the antiphospholipid syndrome, Behcet's disease, …

Other risk factors are **exogenous:** oral contraceptives containing estrogen and pregnancy.

How are the causes diagnosed?

Your doctor will perform blood tests to make a diagnosis. These tests include genetic testing for genetic disorders (thrombophilia) and other tests to diagnose myeloproliferative disorders such as Jak2 V617f/exon 12 or Cal r mutations, and other tests to discard antiphospholipid syndrome (antiphospholipid antibodies, lupus anticoagulant, anti beta2 gp1). He will also discard other more rare causes (see chapter what are the causes). All these tests need to be performed, as sometimes several causes can be identified. Diagnosis of these associated diseases may be more difficult than in the absence of BCS, and deserve exhaustive screening by expert centres.

In the case of myeloproliferative disorder, further tests can be required, including a bone marrow biopsy or a measurement of the blood mass.

How is BCS treated?

The mainstay of treatment of BCS is anticoagulation (to put the blood thinner). In more severe cases and always specific to each patient, stenting, angioplasty in ordert o recanalize the obstructed vein may be done by an interventional radiologist. If the situation is more severe, TIPS (as explained in the text) must be done. Liver transplantation remains the last and unfrequent solution for symptomatic BCS.

At the same time that BCS is treated it is fundamental to recognize what has caused it as specific treatments directed towards myeloproliferative disorders, automimmune diseases, etc must be started. This will also change the prognosis.

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Anticoagulant agents (blood thinners)

These decrease the blood's tendency to clot and prevent future blood clots (thrombus=obstruction) which could block blood flow in the veins, and extension of a preexisting clot. Two kinds of blood thinners are available, the oral ones and those given by injection.

In the case of oral treatment with vitamin K antagonists, regular blood tests are performed to measure INR. *There are new oral blood thinners that do not need to be monitored.*

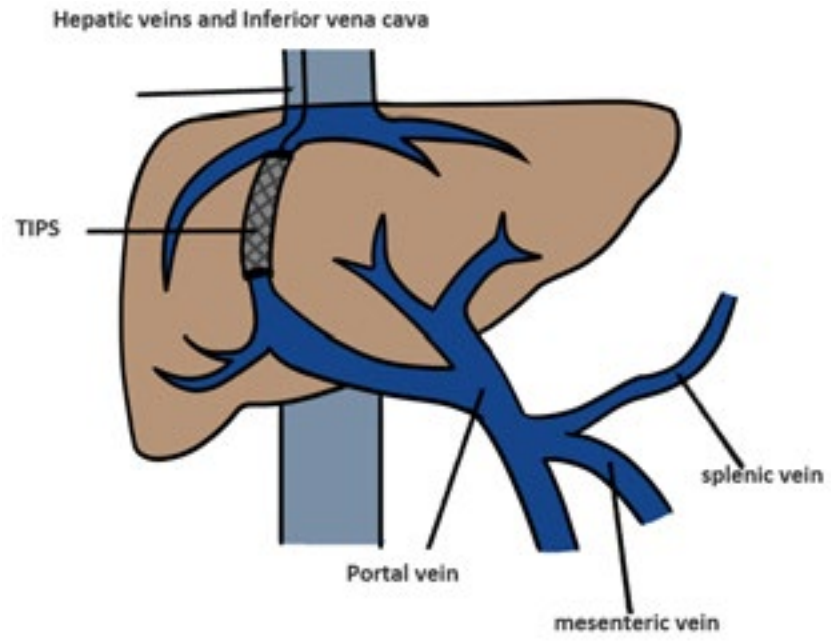
Whatever anticoagulation therapy is chosen, it needs to be given to the patient lifelong.

Radiological procedures

There are two non-surgical procedures used in treating Budd-Chiari syndrome: percutaneous transluminal angioplasty and transjugular intrahepatic portosystemic shunt (TIPS).

Percutaneous transluminal angioplasty procedure, the doctor inserts a catheter (a thin, hollow tube with a balloon at the tip) through the skin and into a blood vessel. The catheter is guided to the area where the clot is located. When the catheter reaches the clot, the balloon is inflated to widen the vein. A stent may be placed at the site to keep the vein open.

Transjugular intrahepatic portosystemic shunt (TIPS) is a radiologic procedure in which a stent (a tubular device) is placed in the middle of the liver to reroute the blood flow. During the procedure, a radiologist makes a tunnel through the liver with a needle, connecting the portal vein (the vein that carries blood from the digestive organs to the liver) to one of the hepatic veins (the three veins that carry blood from the liver). A metal stent (a small, hollow tube) is placed in this tunnel to keep the track open. The TIPS procedure reroutes blood flow in the liver and reduces pressure in all abnormal veins, including the bowel and the liver.



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Transplantation

Liver **transplantation** is considered when patients do not respond to angioplasty or TIPS adequately or in case of very severe acute presentation of the disease.

Therapeutic Strategy

Therapeutic options are implemented in a step-by-step manner. The less invasive option is used first, which is the blood thinners, angioplasty/stenting and/or TIPS. Only when the response is not adequate, **transplantation** is considered. Choosing a more invasive option depends on the clinical response to the previous less invasive one (if it is technically feasible).

Treatment of the cause, when diagnosed, is also very important. Patients often need to be followed by more than one specialist.

Side effects

Complications arising from Budd Chiari syndrome can be related to the disease itself (ascites, variceal bleeding, liver function impairment, and infection). Other complications can appear as a result of certain therapies (bleeding because of blood thinners, or severe side effects resulting from angioplasty or TIPS).

Recurrences of thrombosis

Recurrences are unlikely if prescribed intakes of blood thinners are respected and monitored (INR follow up). To avoid the recurrence of thrombosis, it is essential to follow medical prescriptions. If any problems arise, you should discuss them with your practitioner without delay.

Prognosis

Patients can find the first two years difficult and they require specialized treatments. After this period, the liver condition stops being a problem for 80% of patients. From then on, treatments focus on the cause of the disorder. It is possible to have an almost normal family life and professional life.

Who is affected?

BCS is rare, with an annual incidence between 1-2 persons per 1.000.000. Generally young adults from 20 to 50 years old, with a majority of women. Younger patients or elderly patients are less frequently affected but it may possibly occur.

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What can I do?

Medication

Follow and understand medical prescriptions and inform others of these prescriptions (i.e. with a card in your wallet). Oestroprogestative pills will need to be interrupted, and advice from your gynaecologist is needed. It is also always better to anticipate and prepare for pregnancy in this situation. If this is the case speak to your hepatologist about it, so that a multidisciplinary checkup is performed before, and treatment modified accordingly.

Diet

Avoid alcohol or any co-factor for liver disease, in particular overweight.
Therefore a balanced diet is recommended.

Family planning

BCS can be difficult to manage physically and psychologically. Family support and information are important, especially in the first months after diagnosis.

Pregnancy can be a cause of BCS and can aggravate BCS when the situation is not stabilized. On the other hand, it has been shown that pregnancy is possible, but needs to be anticipated and prepared by medical staff, and family. It should be rapidly discussed to anticipate treatment modifications during pregnancy, and complete work up.

Do my family and I need to get genetically tested?

BCS is not a congenital disease. Nevertheless, as multiple co-factors can be present, it is important to screen for predisposing congenital anomalies or risk factors. These can be tested in children or other family members of patients who have these congenital anomalies.

Do I need medical checkups?

Yes, it is very important to have regular blood tests, imaging and outpatient clinic checkups, at least every 6 months regularly.

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How can I find a specialist?

BCS may need to be managed by a doctor or a hospital network with experience or an interest in BCS.
BCS is a rare disease that not every doctor has an interest in or experience with. However, some hospitals are part of the Europe-wide reference 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/. You can find information and patient support in the section "patients" on the ERN RARE-LIVER website (https://rare-liver.eu/).

Disclaimer

The information provided free of charge on our website has been compiled to the best of our knowledge in order to give interested readers an initial overview of possible diseases and treatment options.
They are intended solely for informational purposes and in no case replace personal advice, examination or diagnosis by authorized doctors.

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