Press release
ERN RARE-LIVER

What is ERN RARE-LIVER?

The European Reference Network for Rare Liver Diseases (ERN RARE-LIVER) is a network of medical centres of excellence initiated by the European Commission in 2017 for the diagnosis and treatment of affected individuals, both children and adults.

The aim is to pool the knowledge and experience of proven experts across Europe and make it available to all patients and their treating physicians. There are 53 university hospitals from 15 EU countries as full members of the network, and an additional 27 centres from 13 other countries are linked to the network through partnerships. All of them work closely together.

In addition to its expertise, the ERN is also characterised by its consideration of patient interests. Thirty-nine different patient organisations are affiliated to the network and send representatives to the Management Board and all other stakeholders of the network.

For people with rare diseases, it is often very difficult to get the right diagnosis and the necessary, often very individual treatment options in time. In the European Union, a disease is considered rare if it affects no more than five in 10,000 people. This corresponds to a disease frequency of less than 0.05 percent, which means that even the average specialist has never seen the corresponding clinical picture, or at most only once. Expertise regarding rare diseases can therefore only be found at a few specialised centres. The aim of ERN RARE-LIVER is to give every affected patient access to this specialised knowledge and to jointly improve the diagnosis and therapy of these rare diseases.

To this end, the network offers extensive information material, organises seminars, workshops and webinars for further education and is represented at international congresses in cooperation with the European societies. Special working groups are dedicated to the various clinical pictures and to overriding topics such as the quality of life of affected patients or the care structures.

As a very important service, the network also offers targeted advice for individual cases via a data-secured online tool. For this purpose, every doctor can contact one of the member centres, and from there the consultation of the best specialists for the respective problem can be initiated easily and quickly. In this way, more than 400 patients with particularly complex problems in rare liver diseases have already been helped.

In this way, the ERN RARE-LIVER network is gradually achieving the overall goal of making the best knowledge about liver disease available to every European patient, regardless of where they live.

Two specific examples:

Autoimmune hepatitis (AIH) is a rare inflammatory disease of the liver in which the patient's own immune system attacks and damages the liver, so AIH could also be called "rheumatism" of the liver. If left untreated, the disease progresses until the liver is so badly damaged that it can no longer fulfil its function and the patients die, unless a liver transplant is still possible. Anti-inflammatory therapy can stop this process, improve the quality of life and patients usually regain a normal life expectancy. A liver transplant is therefore no longer necessary.

The very rare liver disease, Wilson's disease or copper storage disease, has a completely different background. In this case, a genetic (i.e. inherited) defect disturbs the metabolism of copper, on which the body depends in small quantities: copper continues to be absorbed into the body, but no more copper is excreted. Thus, copper accumulates in various organs such as the liver, but also the nervous system, and causes damage. Modern drug therapies that bind copper and promote copper excretion can effectively prevent organ damage.

As effective as the therapy can be, both diseases require individualised adjustment to the respective patient and therefore, experience and specialised knowledge.

How does the patient find the right doctor?

Patients normally go to their family doctor, who often has no idea of the symptoms and clinical picture in the case of rare liver diseases, so he or she refers the patient to the nearest university hospital where further examinations take place. But even in most hospitals, the expertise is often not enough to find out what the right treatment would be. In this case, doctors turn to the European Reference Network on Hepatological Diseases (ERN RARE-LIVER).

Having obtained patient's consent, the hospital enters the file into ERN's highly secure online system. Specialists from all over Europe analyse the case and discuss the possible options. Based on similar cases, the panel advises on a treatment tailored to this individual case, which in most cases can then be carried out in the patient's home country. However, the system can also be used to obtain expert opinions on diagnosis, i.e. in unclear cases, to ask experts what they think the disease is, and to find out what further tests are needed to obtain the diagnosis.

"This means that the patient does not have to travel after the expert knowledge at great expense in terms of time and money; instead, our network brings the knowledge directly to the patient," explains liver specialist and network coordinator, Professor Ansgar W. Lohse.

Background information:

In the European Union, a disease is considered rare if it affects no more than five in 10,000 people. There are many of these diseases. Rare diseases often have a genetic component as their cause. They often run a chronic course and frequently reduce the life expectancy of the patient. Congenital diseases are distinguished from diseases that have infectious or toxic causes.

There are various rare diseases with a genetic cause that can lead to liver damage. These include, for example, Wilson's disease, porphyria, or alpha-1 antitrypsin deficiency, which are caused by defects in enzymes that are necessary for the breakdown of metabolic products.

"Early detection is particularly important in liver diseases. The disease and its consequences can be treated and possibly even cured the earlier they are detected," explains Professor Ansgar W. Lohse.

Functions of the liver

The liver has numerous functions. It plays an important role in energy metabolism and also produces many essential enzymes to regulate it. Its tasks also include detoxifying the body, storing energy reserves and vitamins, and producing blood proteins, bile, defence substances and starting products for hormone production. Important components of the immune system are also located in the liver. These are primarily defence cells that serve to fight off infections, but they are also important for the balance of the immune system throughout the body. A functioning liver is essential for the health of every human being.