The Childhood Liver Disease Research Network strives to provide information and support to individuals and families affected by liver disease through its many research programs.

**CYSTIC FIBROSIS LIVER DISEASE**

**What is Cystic Fibrosis Liver Disease?**

Cystic Fibrosis (CF) is a condition affecting the lungs, pancreas, liver and other organs that are caused by changes (mutations) in a gene called “CFTR” (Cystic Fibrosis Transmembrane Conductance Regulator). The primary problems in most patients with CF are decreased functioning of the pancreas and chronic lung disease. Mild liver involvement with abnormal liver blood tests or fat in the liver is common. Gallstones and very small gallbladder are common. In some patients, the liver problems caused by CF are much more significant than the lung disease. Advanced liver disease in individuals with CF is called CF liver disease (CFLD). Severe scarring of the liver, called cirrhosis, can occur in CFLD.

**What are the symptoms of the Liver Disease in Cystic Fibrosis?**

Many CF patients with liver involvement do not have any symptoms. In advanced CFLD or cirrhosis, patients may have a large liver and spleen. Problems with weight gain can occur. As the scarring in the liver becomes more severe, the pressure in the vein leading to the liver (the portal vein) rises and complications of portal hypertension can occur. These can include a large spleen, fluid accumulation in the abdomen (ascites) and bleeding from dilated veins (varices) in the esophagus or stomach. Rarely jaundice and bleeding problems can occur.

**What is the Lung Disease in Cystic Fibrosis?**

In cystic fibrosis, the salt balance in the body is disturbed. Because there is too little salt and water on the outside of the cells, the thin layer of mucus that helps keep the lungs free of germs becomes very thick and difficult to move. And because it is so hard to cough out, this mucus clogs the airways and lead to infections that damage lungs.

**What Problems Can Cystic Fibrosis Cause in Other Organs?**

*Pancreas:* In about 90% of patients with CF the tubes that drain the pancreas are blocked and the release of the natural enzymes that aid in the digestion of fat and
protein is prevented; this is called "exocrine pancreatic insufficiency." Affected patients need to take pancreatic enzymes with food to help with digestion. Some patients can develop inflammation of the pancreas (pancreatitis).

**Diabetes:** CF related diabetes can occur in individuals with CF beginning in the pre-teenage years

**Meconium ileus:** About 10% of patients with CF are born with thick secretions blocking the lower part of the small intestine called meconium ileus.

Constipation is common in CF. Some patients with CF will develop a separate problem, the onset of bowel blockage call distal intestinal obstruction syndrome (DIOS). While this is different from constipation, the two may be confused.

**How do you get Cystic Fibrosis Liver Disease?**

It is not known why some people with CF develop CFLD and why others do not. It is believed that the role of the CFTR protein is to add liquid to bile and that in CF the bile becomes thick and leads to plugging of the bile ducts resulting in inflammation and scarring.

**How is Cystic Fibrosis Liver Disease found and diagnosed?**

CF liver disease is found during physical exam by a doctor and by blood tests. Sometimes ultrasound, CT scans, liver biopsy and other tests may add information.

**What is the Treatment for Cystic Fibrosis Liver Disease?**

There is no known cure for CFLD, but there are ways to prevent or reduce health problems related the liver disease of CF. Some experts believe that ursodeoxycholic acid can help reduce the plugging of the bile ducts, but that has not been proven. Intestinal bleeding can occur in severe CFLD, and medicines and endoscopic or radiologic procedures can be used to treat this. Patients with CFLD should be immunized against hepatitis A and B to prevent further injury to the liver. They should avoid alcohol use. In some cases the liver disease is so severe that liver transplantation may be considered.
What is the Outlook for someone with CF Liver Disease?

Most patients with CFLD can lead long and productive lives. However, those with severe lung problems or progressive liver disease may have a shortened life span. Sometimes, if other organ systems are healthy, liver transplant can be done.

Does the ChiLDReN Network have any studies that include patients with CF Liver Disease?

Yes. The ChiLDReN Network currently has one study that includes patients with CFLD. The PUSH study is a natural history study that includes patients with CFLD. A natural history study is aimed at acquiring information and data that will provide a better understanding of rare conditions. Participants will be asked to allow study personnel to obtain information from medical records and an interview, and to collect blood, urine, and tissue samples when clinically indicated, in order to understand the cause of this disease and to improve the diagnosis and treatment of children with this disease. All of the information obtained in this study is confidential and no names or identifying information are used in the study.

PUSH: A longitudinal study of the risk of hepatic cirrhosis in Cystic Fibrosis.
Eligibility: Children ages 3 through 12 years of age with Cystic Fibrosis and pancreatic insufficiency who are enrolled in the CFF or Toronto CF registry studies. This study is closed to enrollment.
ClinicalTrials.gov Study NCT01144507

Are there any organizations or foundations that help families dealing with CFLD?

Yes. The ChiLDReN Network works with numerous groups that support patients and families who are dealing with rare liver diseases. Please click here to go to that page on our website (Information for Families). You will see the list of groups and information about them.