



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

FREQUENTLY ASKED QUESTIONS

- The Liver and What it Does
- Cholestatic Jaundice
- Caring for a Child with Cholestatic Liver Disease
- Genetics and Cholestatic Liver Disease

The Liver and What it Does

What is the liver?

The liver is the largest organ in the body. It is essential in keeping the body functioning properly. It removes poisons from the blood, produces agents to control infection, and removes germs and bacteria from the blood. It makes proteins that regulate blood clotting and produces bile to help absorb fats and fat-soluble vitamins.

What are signs of infant liver disease?

In a baby there can be one or more signs that the liver is not working properly. The skin and eyes may be jaundiced. The abdomen may look swollen or stick out. The urine may be dark yellow or brown. The stools are often grey or white instead of green or yellow. There may also be bleeding or easy bruising. The blood might contain higher than normal levels of liver enzymes. The liver may feel large or look large on an x-ray. In infants, Cholestatic jaundice is different from psychologic jaundice.

Cholestatic jaundice in infants requires further testing.

Cholestatic Jaundice

What is cholestatic jaundice?

Cholestatic jaundice results when there is a blockage of bile flow. This can happen for many different reasons. It may result from a brief illness or from injury to the liver; when these are the causes it will frequently go away without any special treatment. It can be



caused by a number of inherited conditions (metabolic disease) that may need treatment. It can be caused by drugs and often goes away when the drug is stopped. Cholestatic jaundice can also be the result of more serious liver diseases such as idiopathic neonatal hepatitis or biliary atresia.

How is cholestasis diagnosed?

Cholestasis is diagnosed by blood tests. Usually there is an elevation of conjugated (direct) bilirubin in the blood or elevated bile salts in the blood. To determine the cause of the may involve a number of other blood and urine tests, x-ray or ultrasound tests of the liver and bile ducts, or a liver biopsy.

What are the types of cholestatic liver disease?

- [Biliary atresia](#)
- [Idiopathic neonatal hepatitis](#)
- [Alagille syndrome](#)
- [Alpha-1 antitrypsin deficiency \(Alpha-1\)](#)
- [Bile acid synthesis defect](#)
- [Progressive familial intrahepatic cholestasis \(PFIC\)](#)
- [Mitochondrial hepatopathy](#)
- [Cystic Fibrosis Liver Disease](#)

Caring for a Child with Cholestatic Liver Disease

Do most insurance plans cover the treatments for cholestatic liver diseases?

Yes, they usually cover the general health needs of children with these diseases although certain specific treatments for rare diseases may not be covered.

Will my child ever grow out of this disorder?

There are some mild forms of many of the disorders that children may outgrow. However, many of the disorders are lifelong and some may even lead to the need for liver transplant.

Does my child need a specialist or can my pediatrician treat this disorder?



Children with cholestatic liver diseases are usually best cared for by a specialist (pediatric hepatologist) who can work with your pediatrician to provide the best care for your child.

We live in a community where we only have access to a small community hospital. Will the medical staff be able to care for my child if he has a crisis? What should we do?

The best way for you to help your child is to seek a specialist early in the course of your child's disease who can work with your pediatrician or family doctor.

My child has just been diagnosed with cholestatic liver disease. Will my child be able to live a normal life?

Early in the course of your child's disease it is usually not possible to predict the future with certainty. Your hepatologist will provide you with the most specific information about the future for your child depending on the cause and severity of the cholestatic liver disease.

Do children with cholestatic liver diseases need a special diet?

Children with these diseases often need special formulas which can be easily absorbed and they may require extra vitamins particularly vitamins A, D, E, and K.

Can children with a cholestatic liver disease participate in regular physical activity?

The jaundice itself does not mean that your child should be excluded from regular physical activity. However, sometimes children with cholestatic liver diseases develop big spleens and should not participate in contact sports. Also, these children may have brittle bones which can easily be broken. For these reasons, it may be recommended that your child avoid contact sports.

We have a child with cholestatic liver disease. Will we be able to care for our child ourselves? Will my child be able to go to school?

Depending upon the specific cause of the cholestatic liver disease, your child may need specialized nursing care. One such example would be if he or she were to have a liver



transplant. Your hepatologist will make every effort to help your child function as normally as possible, including attending school.

What is the life expectancy of a child with a cholestatic liver disease?

The answer to this question depends a great deal on the specific diagnosis.

Why isn't there a cure or better treatment for cholestatic liver diseases?

There are currently several treatments for the symptoms of some cholestatic liver diseases including medications to treat itching.

While treatments for symptoms associated with cholestatic liver disease have improved over the years, much remains to be learned. These liver diseases are rare and, in the past, it has been difficult for pediatric liver specialists to find funding to support studies of these diseases. However, our network, ChiLDREN (Childhood Liver Disease Research and Education Network) which is supported by the National Institutes of Health, will conduct research on the causes and treatment of these diseases in order to provide new hope for children with cholestatic liver disease.

Will my child ever grow out of this disorder?

Since these disorders are hereditary or genetic, the child will not outgrow his/her disorder. However, several of these disorders appear to improve as children age, and with optimal care and treatment many of these children will improve greatly.

Genetics and Cholestatic Liver Disease

My child has been diagnosed with cholestatic liver disease. Are my other children at risk and should they be tested?

To the best of our knowledge, neither biliary atresia nor idiopathic neonatal hepatitis is a genetic inherited disease and your other children are not at risk. However, the genetic diseases being studied include: Alpha-1 anti-trypsin deficiency, Alagille syndrome, progressive familial intrahepatic cholestasis (PFIC), bile acid synthesis defect (BAD), and mitochondrial hepatopathies.

We have a child with a cholestatic liver disease and want to have more children.



Will subsequent pregnancies be affected? Is prenatal testing available?

Please talk to your hepatologist about your child's specific disease for the most accurate answer to these questions.

Is DNA testing available and/or necessary to diagnose the disorders?

Clinical DNA testing is available for Alpha-1, Alagille syndrome, PFIC and mitochondrial hepatopathies.

Will my child pass this disease on to his/her children?

Your child would not pass biliary atresia or idiopathic neonatal hepatitis on to his/her children. Please talk to your pediatric liver specialist about your child's specific disease for the most accurate answer to these questions.

Are any specific ethnic groups affected by cholestatic liver diseases?

No.