Alagille Syndrome (paucity of bile ducts, hyperbilirubinemia)

Patient and family information, brought to you by the Education Committee of APSA

Overview - "What is it?"

Alagille syndrome is a type of disease in which the baby has several abnormalities. In Alagille syndrome, one of the main problems is that there are fewer than the normal number of bile ducts in the liver. The liver cells make bile, a yellow liquid that the body needs to help digest food that a person eats. The bile is transferred from the liver cells into the intestine through bile ducts. The bile ducts start out small in the liver, then join together like smaller twigs that join into larger and larger branches making up the main bile ducts or channels that bring the bile from the liver into the intestine. In Alagille's syndrome where there is a fewer number of bile ducts, the bile gets backed up in the liver (See Figure 1). The baby's skin and the whites of the eyes can turn yellow (jaundice).

Children with Alagille syndrome may also have other abnormalities such as heart defects, abnormal vertebrae/spine bones, a lung artery abnormality, characteristic facial features, and an enlarged liver.

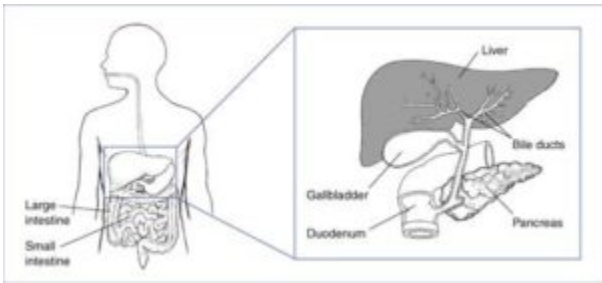

Figure 1: Structure of the liver and the bile ducts

Patients with Alagille Syndrome have fewer bile ducts to drain bile. *National Institute of Diabetes and Digestive and Kidney Diseases, National Institutes of Health.*

An estimated 1 in every 70,000 children born will have Alagille syndrome. This is a genetic disorder in which there is an abnormality in a gene which is passed onto the child by the parent.

Signs and Symptoms - "What symptoms will my child have?"

Early signs: Because of the bile duct problem, the baby may have yellowing of the whites of the eyes and skin. As the baby gets older, the liver can get very big and does not work well. With poor liver function, the child can have belly pain, itching, dark (brown) urine and light (grey-colored) stool. The child may have poor appetite, feel queasy or sick resulting in poor growth/weight gain.

The facial features that may be seen are shown below. The face can have a broad, prominent forehead, deep-set eyes, and a small, pointed chin. (Figure 2)

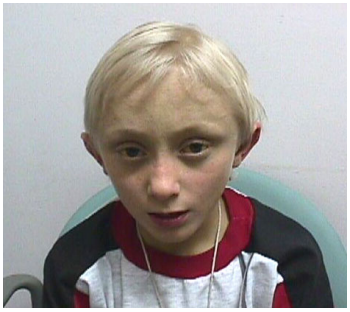

Figure 2: Common Facial Features of Alagille Syndrome
Photo courtesy of: The Childhood Liver Disease Research Network

Diagnosis - "What tests are done to find out what my child has?"

Labs and tests:

Bloodwork to measure the function of the liver and the level of bile in the blood stream.

Ultrasound is a test that can look at the liver and the gallbladder to seek other reasons that can cause jaundice such as stones in the gallbladder, abnormality of the large bile ducts, or a tumor of the liver.

HIDA scan is a nuclear medicine test that looks to see if bile from the liver makes it into the intestine.

Eventually, the child may need to have surgery to biopsy the liver and to inject some dye into the bile ducts to see if there is a blockage. Most children will also get a heart ultrasound and X-rays of the spine to help with the diagnosis. There are also eye abnormalities that are seen only in Alagille syndrome, so an eye specialist may need to look at the patient's eyes.

Conditions that mimic this condition: Biliary atresia, viral hepatitis, cystic fibrosis, hypothyroidism, alpha-1 antitrypsin deficiency.

Treatment - "What will be done to make my child better?"

Medicine:

- Standard immunizations
- Correction of vitamin deficiency. Some vitamins are not absorbed when the liver is not working well (vitamin A, D, K, E)
- Medicines to treat itching (Diphenhydramine, hydroxyzine, or cholestyramine)

Dietary modifications: High carbohydrates and medium chain fatty acids- your child's dietician and doctors will tell you what foods are best

Surgery: Most children will undergo a liver biopsy to confirm the diagnosis. A biopsy is when a piece of liver is taken and studied to see what is wrong with it. Most children will also need a cholangiogram. A cholangiogram is a procedure where dye is injected into the gallbladder to outline the bile ducts from the liver to the intestine. Both procedures (biopsy, cholangiogram) can be done through the skin using needles or through a larger cut in the abdomen. If Biliary atresia is found, this abnormality may be corrected with a surgery, it is really important to rule this out.

In children or adults with progressive failure of the liver, a liver transplant may be needed.

Preoperative preparation: For liver biopsy and cholangiogram, the child will need to have an empty stomach in order to undergo anesthesia. Patients are asked to stop eating or drinking for 8 hours before surgery.

In children needing a liver transplant, a transplant team will make sure that the appropriate preparations are made for the surgery. Your family will be part of this preparation.

Informed consent: A consent form is a legal document that states the tests, treatments or procedures that your child may need and the doctor or practitioner that will perform them. Before surgery, your doctor should tell you what the operation is, the goal of the surgery and other possible treatment options that are available. Your doctor should explain the risks and benefits of the surgery. You give your permission when you sign the consent form. You can have someone sign this form for you if you are not able to sign it. You have the right to understand your child's medical care in words you know. Before you sign the consent form, make sure all of your questions are answered. It is important to know that during surgery, there are things that can happen that your doctor may have not predicted before going in. He or she will explain these to you after the surgery.

Emotional support: Stay with your child for comfort and support as often as possible while he or she is in the hospital. Ask another family member or someone close to the family to stay with your child when you cannot be there. Bring items from home that will comfort your child, such as a favorite blanket or toy.

Postoperative care: After liver biopsy and cholangiogram, the child will be closely monitored for any type of bleeding from the liver biopsy. Feeding may be restarted shortly after the procedure.

Risks/Benefits: The biggest risk of taking a piece of liver for a biopsy is the bleeding. Other risks include infection, injury to organs near the liver or need for repeat biopsy if the specimen was not good enough. The benefit is getting an accurate diagnosis so that treatment can be started right away.

In some patients, the liver may get so bad that it may not work anymore. When this happens, a liver transplant may be needed. After a liver transplant, the child will be in the intensive care unit (ICU). The transplant team will help with recovery process. The patient will need medicine to keep the body from rejecting the new liver (immune system blockers). Children that undergo a transplant will have to be followed closely for signs of infection, since their immune system will be blocked. They need to be monitored for possibility of rejection.

Home Care - "What do I need to do once my child goes home?"

Diet: A special baby formula may be recommended to support a healthy liver. The baby will likely need vitamins (A, D, E, K).

Activity: Activity can return to normal when the incisions are well healed.

Wound care: For biopsy and/or cholangiogram, leave the wound dry for about five days, then the baby can bathe. If the patient underwent liver transplant surgery, the surgical team will let you know when it is safe to given them a bath. Contact your surgeon for any redness or drainage of fluid around the incision or if your child has any fever.

Medicines: Your child may need medicines to help with itching and to help with allowing the bile to travel from the liver to the intestine more easily. Medicines for pain may be needed including oral narcotics. Stool softeners and laxatives are needed to encourage regular bowel movements after surgery, especially if narcotics are still needed for pain.

What to call the doctor for: Call your doctor for uncontrollable itching, vomiting, abdominal pain, fever, or any problems with the incisions.

Follow-up care: You should follow up with your pediatrician to monitor growth and development. Schedule appointments to see the gastroenterologist (GI specialist) and the surgeon within a few weeks after returning home from the surgery. Follow up with a pediatric cardiologist (heart doctor) may also be important.

Long Term Outcomes - “Are there future conditions to worry about?”

Children may develop worsening liver disease and eventually require a liver transplant. Growth and development are often affected, so physical and occupational therapy are helpful. Cardiac and liver disease can affect the lifespan of the child, so follow-up with a pediatric cardiologist and gastroenterologist is extremely important.

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