



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.

## TERM GLOSSARY

### A

**Abdomen:** The portion of the body between the chest area and the pelvic area (hips). Includes the stomach, intestines, liver, spleen, pancreas, kidneys, appendix, bladder, gallbladder, urinary bladder and other parts of the digestive system.

**Abnormal Mitochondrial DNA:** The way nutrients from food move from the small intestine into the cells in the body.

**Absorption:** A change from normal in the code of the DNA that is present in mitochondria in each cell.

**Acholic:** a lack or absence of bile secretion. Acholic stools appear pale or grayish in color.

**Actigall:** a man-made bile acid used in treating liver diseases. See also Ursodeoxycholic Acid

**Acute:** Having a rapid onset and a short course.

**Acute Liver Failure:** Abrupt onset of a serious disease causing failure of the liver to perform its normal functions

**Alpha-1-antitrypsin:** A normal protein which functions to regulate protein breakdown by the enzyme trypsin. When alpha-1-antitrypsin is deficient in the bloodstream because of a defect leading to excess accumulation of alpha-1-antitrypsin in the liver and lungs, these organs may be damaged. Please see the Information for Families section for more information on alpha-1-antitrypsin liver disease.

**Amish:** A religious group of people, typically located in rural, farming areas. They are known for their rejection of typical modern-day conveniences such as electricity and motor vehicles.

**Ammonia:** A chemical in the blood that is normally removed by the liver.

**Anastomosis** (AN-nah-stuh-MOH-sis): An operation to connect two body parts. An example is an operation in which a part of the small intestine is removed and the two remaining ends are rejoined.

**Anemia:** A condition in which there is a low number of red cells in the blood.

**Antihistamines:** Drugs that counter the effects of histamine, a normal chemical in the blood that is believed to cause allergic reactions.

**Aorta:** The main artery coming from the heart and taking blood to the rest of the body.



**Appetite:** Desire for food.

**Ascites** (uh-SY-teez): A buildup of fluid in the abdomen. Ascites is usually caused by severe liver disease such as cirrhosis.\*

**ATP:** A chemical made in the mitochondria which stores and delivers energy.

**Atresia** (uh TREEZ-ya): Lack of a normal body opening such as a bile duct.

**Autosomal Recessive:** Refers to a gene for a specific characteristic (such as blue eyes rather than brown eyes) that will not show up unless a baby has inherited the recessive (also known as non-dominant) gene from both parents.

## B

**Benign:** Not likely to become progressively worse or cause death; not recurrent; usually meaning favorable for recovery.

**Bile:** A clear yellow or orange fluid made by the liver and stored in the gallbladder. Bile helps break down fats and gets rid of waste in the body. Bile is passed to the small intestine for use in digestion.

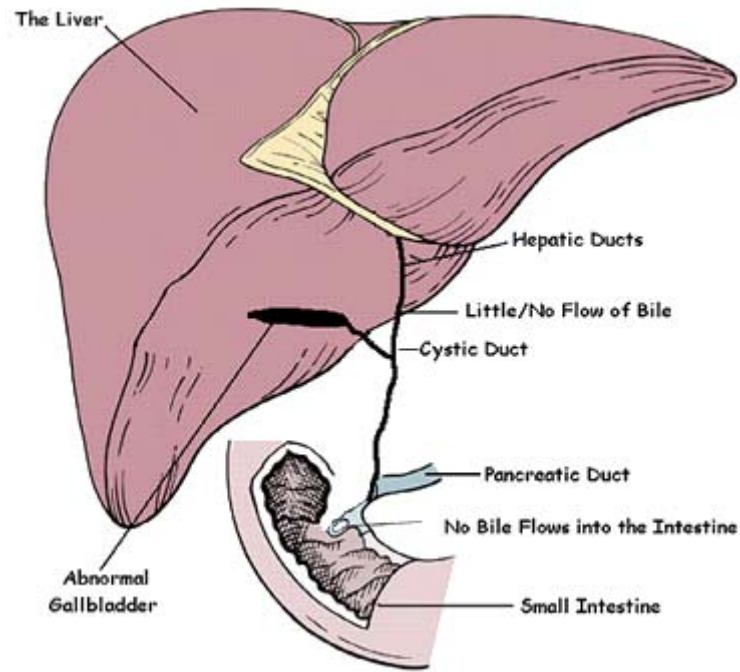
**Bile Acids:** Steroid acids that come from cholesterol and are broken down in the liver.

**Bile Ducts:** The passageways through which bile moves. There are three bile ducts which connect the liver to the large intestine and pancreas.

**Bile Salts:** A component of bile that helps to break down fats so they can be properly used by the body.

**Biliary Atresia:** A condition in which the bile ducts inside or outside the liver do not have normal openings. Bile becomes trapped in the liver, causing jaundice and cirrhosis. The gallbladder may be missing or abnormal. Without surgery the condition may cause death. (see Figure 1 below).

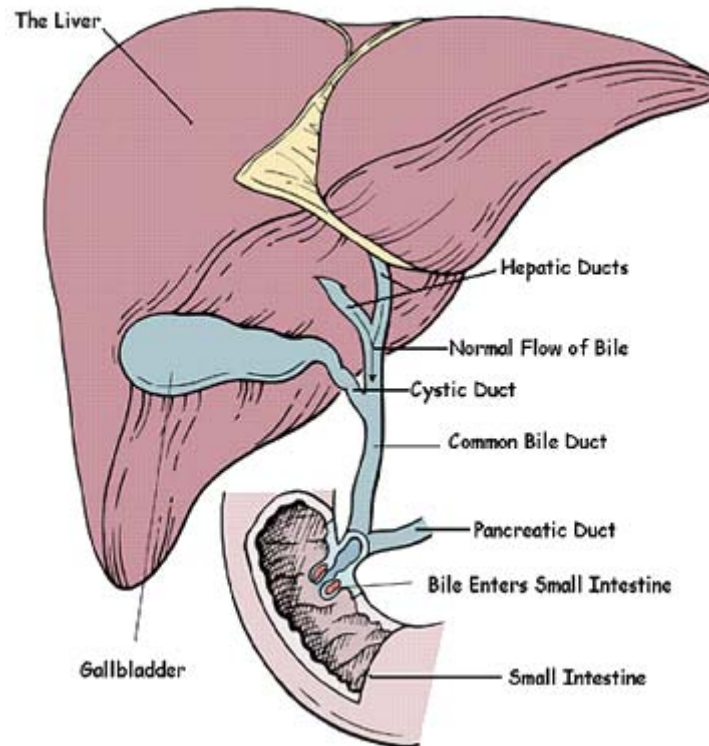
**Figure 1: Biliary Atresia**



**Biliary stricture:** A narrowing of the biliary tract from scar tissue. The scar tissue can result from injury, disease, or infection.

**Biliary tract:** The gallbladder and the bile ducts. Also called biliary system or biliary tree. (see Figure 2 below)

**Figure 2: Biliary Tract**



**Bilirubin:** The substance formed when hemoglobin breaks down. Bilirubin gives bile its color. Bilirubin is normally passed in stool. Too much bilirubin causes jaundice.\*

**Biopsy:** Removal and examination of tissue from a living person; usually used to determine whether a tumor is life-threatening or benign.

**Blood Clot:** A clotted mass of blood.

**Blood clotting:** The formation of a jelly-like substance within blood vessels that stops blood from flowing.

**Bowel:** Another word for the small and large intestines.

**Butterfly vertebrae:** A clefting abnormality in small bones that make up the spine that gives them a "flying butterfly" appearance on X-rays.

**Byler's Disease:** A disease typically associated with Amish children suffering from Progressive Familial Intrahepatic Cholestasis.



## C

**Cancer:** A group of diseases in which normal body cells begin to grow abnormally, resulting in tumors.

**Carriers:** A person who has genes for a specific disease, but does not develop symptoms of that disease.

**Cholangiography** (koh-LAN-jee-AW-gruh-fee): A series of x-rays of the bile ducts.

**Cholangitis** (KOH-lan-JY-tis): Irritated or infected bile ducts.

**Cholecystectomy** (KOH-lee-sis-TEK-tuh-mee): An operation to remove the gallbladder.

**Cholecystitis** (KOH-lee-sis-TY-tis): An irritated gallbladder

**Cholestasis:** A group of diseases characterized by poor bile flow and build-up of substances in the liver that should be put into bile.

**Cholestatic jaundice:** yellowing of the skin resulting from abnormal bile flow in the liver.

**Cholesterol:** A fat-like substance that is made by the body and is found naturally in animal foods such as meat, fish, poultry, eggs, and dairy products. Foods high in cholesterol include liver and organ meats, egg yolks, and dairy fats.\*

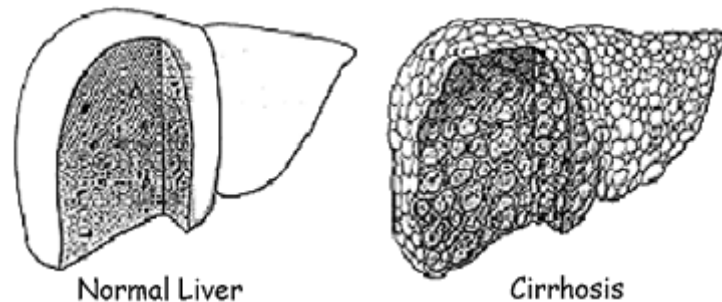
**Cholic Acid:** An acid formed in the liver from cholesterol. Along with other bile acids, cholic acid is important for digestion.

**Chromosomes:** Found in all human body cells, chromosomes contain DNA (genetic information) which determines individual characteristics, such as whether a person will have blue eyes or brown eyes, or whether someone might get liver disease.

**Chronic:** Persisting for a long time; referring to a disease that shows little change or improvement or is expected to last a long time.

**Cirrhosis:** A disease of the liver in which scar tissue replaces normal, healthy tissue, blocking the flow of blood through the organ and preventing it from working as it should. \*\* Cirrhosis makes it hard for the liver to remove poisons (toxins) from the blood. These toxins build up in the blood and may affect brain function. (see Figure 3 below)

**Figure 3: Cirrhosis**



**Colon:** A small portion of the large intestine, just before the rectum.

**Common bile duct:** The tube that carries bile from the liver to the small intestine.

**Common Bile Duct Obstruction:** A blockage of the common bile duct.

**Congenital Heart Disease:** Heart disease that is present at birth.

**Constipation:** Constipation is passage of small amounts of hard, dry bowel movements, usually fewer than three times a week. People who are constipated may find it difficult and painful to have a bowel movement. Other symptoms of constipation include feeling bloated, uncomfortable, and sluggish.\*\*

**Convulsions:** Fits or seizures caused by abnormal electrical activity in the brain.

**CT/CAT Scans:** Computerized Axial Tomography. Similar to an x-ray, this test takes pictures of small, specific portions of the inside of the body.

**Cystic duct:** The tube that carries bile from the gallbladder into the common bile duct and the small intestine.

**Cystic fibrosis:** Cystic fibrosis is an inherited chronic disease that affects the lungs and digestive system of about 30,000 children and adults in the United States (70,000 worldwide). A defective gene and its protein product cause the body to produce unusually thick, sticky mucus that clogs the lungs and leads to life-threatening lung infections and obstructs the pancreas and stops natural enzymes from helping the body break down and absorb food.

**Cystic fibrosis liver disease (CFLD):** Bile secreted by the liver to aid in digestion may block the bile ducts, leading to liver damage. Over time, this can lead to scarring and cirrhosis. The liver fails to rid the blood of toxins and does not make important proteins such as those responsible for blood clotting.

**D**

**Deficiencies:** A lack or shortage of something needed by the body to function properly.

**Diagnosis:** A determination of the nature of an illness or disease. A diagnosis will also usually describe the cause and expected outcomes of a disease.

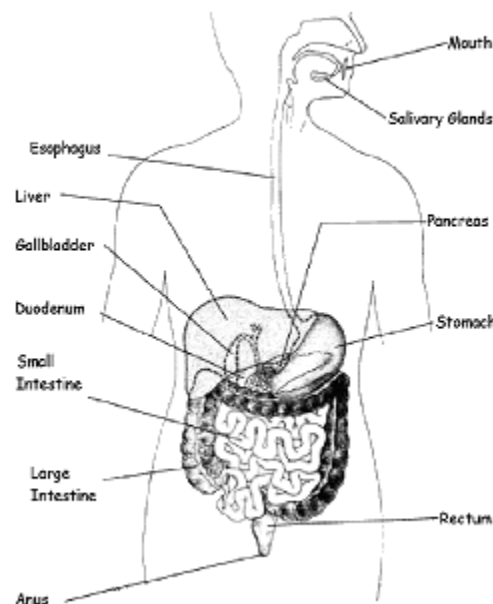
**Diagnostic Tests:** Medical tests, such as examination of blood or visual exam, to assist medical personnel in determining the cause of illness, injury or disease.

**Diarrhea:** Rapid movement of feces through the intestine, causing poor absorption of water and nutrients, as well as watery stools.

**Digestion:** The process the body uses to break down food into simple substances for energy, growth, and cell repair.

**Digestive System:** The organs in the body that break down and absorb food. Organs that make up the digestive system are the mouth, esophagus, stomach, small intestine, large intestine, rectum, and anus. Organs that help with digestion but are not part of the digestive tract are the tongue, glands in the mouth that make saliva, pancreas, liver, and gallbladder. (see Figure 4 below)

**Figure 4:** Digestive System



**Disabling:** Causing an inability to function normally.

**Distention:** Bloating or swelling of the abdomen.

**Diverted:** To move something in a different direction, away from the normal path.



## E

**Echocardiogram (ECHO):** a test that uses sound waves to create a moving picture of the heart. The picture is much more detailed than a plain x-ray image and involves no radiation exposure.

**Endoscopic Retrograde Cholangiopancreatography (ERCP)** (en-doh-SKAH-pik REH-troh-grayd koh-LAN-jee-oh-PANG-kree-uh-TAH-gruh-fee): a test where a dye is injected into the bile and pancreatic ducts using a flexible, video endoscope. Then x-rays are taken to outline the bile ducts and pancreas.

**Enteral Nutrition:** A way to provide food through a tube placed in the nose, the stomach, or the small intestine. A tube in the nose is called a NG tube. A tube that goes through the skin into the stomach is called a gastrostomy or PEG. A tube into the small intestine is called jejunostomy or (PEJ) tube. Also called tube feeding.

**Enteritis:** An irritation of the small intestine.

**Enzymes:** Proteins that cause or promote chemical reactions in the body. There are about 10,000 different enzymes in the human body that each have specific duties in helping the body to function properly.

**Excretory:** Referring to material that is excreted or removed.

**Extrahepatic Biliary Tree:** The bile ducts located outside the liver.

## F

**Failure to Thrive:** A condition that occurs when a baby does not grow normally.

**Familial:** Referring to a family unit, as with diseases that likely to occur in more than one family member by chance.

**Fat-soluble vitamins:** are vitamins A, D, E and K. they need bile for absorption by the intestine and absorbed in a fashion similar to dietary fats. When a patient is jaundiced there may be less bile in the intestine to absorb those vitamins and frequently jaundiced patients require additional fat soluble vitamins to avoid becoming deficient in them.

**Fatty Acid Oxidation Defects:** A group of inherited diseases in which fats are not broken down normally to make energy.

**Fetal:** Relating to an unborn child that is anywhere from about 8 weeks after conception to birth.

**Floppiness (Low muscle tone):** Loss of muscle tone; difficulty moving muscles or lifting the head.

**Fretful:** Upset or unhappy.



## G

**Gallbladder:** The organ that stores the bile made in the liver. It is connected to the liver by bile ducts. Eating signals the gallbladder to empty the bile through the bile ducts to help digest fats.

**Gallbladder Bile:** Bile that is found in the gall bladder.

**Gallstones:** A stone-like mass of tissue, called a calculus, that forms in the gallbladder and can cause abdominal pain.

**Gastroenterologist** (GAH-stroh-en-tuh-RAW-lih-jist): A doctor who specializes in digestive diseases.

**Gene Testing:** Testing that is done with the purpose of determining if the person being tested carries a specific gene, such as one that can cause a specific disease.

**Genes:** Genes are a part of chromosomes. They carry all the information about characteristics (also known as "traits") that babies will inherit from their parents, such as eye color or the potential to develop diseases.

**Genetic:** Passed on from parent to child at birth.

**Glucose:** The major sugar made and used in the human body.

## H

**Heart Murmur:** Any sound in the region of the heart other than normal heart sounds.

**Hepatitis:** Inflammation of the liver that sometimes cause permanent damage. Hepatitis may be caused by viruses or by medicines.

**Hepatobiliary Iminodiacetic Acid Scan (HIDA):** An imaging procedure that helps a doctor track the production and flow of bile from the liver to the small intestine.

**Hepatocyte:** Human body cells from the liver.

**Hepatologist (HEH-puh-TAW-luh-jist):** A doctor who specializes in liver diseases.

**Hepatomegaly:** an enlarged liver.

**Hepatopathy:** A condition of the liver

**Hepatoportoenterostomy (HEH-puh-TAW-poor-tow En-tuh-RAW-stuh-mee):** See Kasai Procedure



**Herbal:** Refers to vitamins or other supplements made from plants for the purpose of preventing or treating disease.

**Hyperbilirubinemia:** Too much bilirubin in the blood. Symptoms include jaundice. This condition occurs when the liver does not work normally

**H2-Blockers:** Medicines that reduce the amount of acid the stomach produces. Prescription H2-blockers are cimetidine (Tagamet), famotidine (Pepcid), and ranitidine (Zantac).

## I

**Immunosuppression:** Reduced activation or efficacy of the immune system through drugs or radiation.

**Inborn Error:** Inherited or acquired before birth.

**Infections:** The invasion of an organism or toxin in the body that causes injury or harm to the surrounding area.

**Inherit:** To receive from one's parents by genetic transmission.

**Intestinal Flora:** The bacteria, yeasts, and fungi that grow normally in the intestines.

**Intestinal Mucosa:** The surface lining of the intestines where the cells absorb nutrients.

**Intestines:** A section of tubes, including both the small and large intestine, extending from the end of the stomach to the anus.

**Intrahepatic:** Within the liver.

**Intravenous:** Within a vein, such as in the use of a needle in the vein to deliver medication.

## J

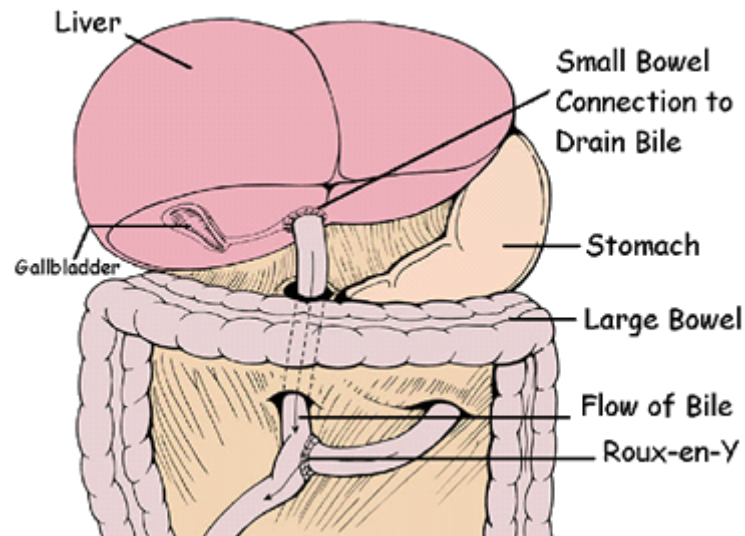
**Jaundice:** A symptom of many disorders. Jaundice causes the skin and eyes to turn yellow from too much bilirubin in the blood.\* See also Hyperbilirubinemia

## K

**Kasai Procedure (KA-sigh):** A surgery performed on an infant with Biliary Atresia to allow bile to flow from the liver. In a Kasai the damaged ducts are removed and replaced with some of the infant's own intestine. The small intestine is divided (Roux-en-Y) and a section is brought up to the liver. This

connection may be inside or outside of the liver. The Kasai procedure is also called a hepatportoenterostomy. (see Figure 5 below)

**Figure 5:** Kasai Procedure



**Ketone Bodies:** A chemical made in the body when a person is fasting (not eating).

## L

**Lactic Acid:** A chemical made in the body in high amounts in respiratory chain disorders.

**Large Intestine:** The part of the intestine that goes from the cecum to the rectum. The large intestine absorbs water from stool and changes it from a liquid to a solid form. The large intestine is 5 feet long and includes the appendix, cecum, colon, and rectum. Also called colon.

**Lipids:** A group of substances comprising fatty, greasy, silky, and waxy compounds that do not dissolve in water. Lipids are a source of fuel for the body.

**Liver:** The liver, the largest organ in the body, is essential in keeping the body functioning properly. It removes or neutralizes poisons from the blood, produces immune 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. You cannot live without a functioning liver.\*\*

**Liver Enzyme Tests:** Blood tests that look at how well the liver and biliary system are working. Also called liver function tests.

**Loose Stools:** Diarrhea.



## M

**Magnetic Resonance Cholangiopancreatography (MRCP):** uses magnetic resonance imaging to get pictures of the bile ducts, pancreatic duct, pancreas, liver and gallbladder.

**Magnetic Resonance Imaging (MRI) Test:** uses a machine that takes pictures of internal structures in the body to determine how they are functioning.

**Malabsorption Syndromes:** Conditions that happen when the small intestine cannot absorb nutrients from foods.

**Malnutrition:** Poor or inadequate nourishment caused by poor diet or a defect in metabolism that prevents the body from using food properly.

**Medium Chain Triglycerides:** A special kind of dietary fat that does not require bile for absorption.

**Microscope:** A machine used to look at human tissues under magnification.

**Mitochondria:** The part of each cell that makes energy for the cell.

**Motor Skills:** A measure of the ability of children and adults to move and have normal strength

**MRI Test:** Magnetic Resonance Imaging testing uses a machine that takes pictures of internal structures in the body to determine how they are functioning.

## N

**Neonatal Hepatitis:** Irritation of the liver with no known cause. Occurs in newborn babies. Symptoms include jaundice and liver cell changes.

**Neonatal iron storage disease (hemochromatosis):** Rapid iron buildup in the liver of a newborn. Left untreated, it can be fatal.

**Nerves:** Cord-like structures in the body that carry information and sensations (like heat, cold and pain) between the brain and other parts of the body.

**Nucleus:** The part of each cell that contains genetic material that directs the actions of the cell.

## O

**Obstruction:** A blockage in the small or large intestine that prevents the flow of liquids or solids.

**Off-Label Indication:** A way of using a drug or medicine that is not typical. For example, in recent years, the use of baby aspirin has become popular in preventing heart disease, although it was developed ("indicated") for easing pain in children.



## P

**Pale Stools:** Bodily waste that is not the normal brownish color, but is greyish or whitish. The pale color can indicate a lack of bile and poor liver function.

**Pancreas:** A large, elongated gland located behind the stomach, between and spleen and the large intestine. It helps the body break down proteins, carbohydrates and fats, but is more typically known for producing insulin. Without insulin, the body develops a disease called diabetes.

**Parenteral Nutrition:** A way to provide a liquid food mixture through a special intravenous tube. Also called hyperalimentation or total parenteral nutrition (TPN).

**Partial Cutaneous Biliary Diversion:** A surgical procedure that diverts some of the bile that flows from the liver through a piece of intestine into a bag attached to the skin.

**Pathologist:** A doctor who specializes in examining tissue samples to make a diagnosis.

**Paucity:** A reduced number of bile ducts that carry bile from the liver.

**Pediatric:** Referring to the branch of medicine that cares for children.

**Peritoneum:** The lining of the abdominal cavity.

**Peritonitis:** Infection of the peritoneum.

**Peroxisome:** A microbody found in vertebrate animal cells, especially liver and kidney cells, that contains urate oxidase and other enzymes.

**PFIC (Prolonged Familial Intrahepatic Cholestasis):** A term to describe a family of diseases characterized by jaundice, itching, and malabsorption of the fat-soluble vitamins. Please see the Information for Families section for more information on the PFIC diseases.

**Phenotype Test:** A blood test that detects differences in alpha-1-antitrypsin that are associated with the deficiency state.

**Phospholipids:** Any lipid that contains phosphorus. They are the major lipids in cell membranes.

**Physiologic jaundice (neonatal jaundice or neonatal hyperbilirubinemia):** Yellowing of the skin and eyes. This is normal in newborn babies, due to the immaturity of the newborn's liver.

**Pigment:** Any coloring matter of the body. A stain or dye.

**Portal Hypertension:** High blood pressure in the portal vein, which carries blood into the liver. This is a common complication of cirrhosis.

**Portal Vein:** The large vein that carries blood from the intestines and spleen to the liver.

**Protein:** A large, organic compound made of amino acid chains joined by peptide linkages.



**Pruritus:** Itching. Pruritus is common in many skin disorders, but also with diseases like diabetes and liver disorders.

**Puberty:** The period of life during which the secondary sex characteristics begin to develop and the body becomes capable of sexual reproduction. Puberty usually occurs between ages 11 and 13 for girls, and ages 13-15 for boys.

**Pulmonary Arteries:** The main arteries leading from the heart to the lungs.

**Pyruvate:** A chemical made in the body.

## R

**Radiologist:** A physician who specializes in radiology, which is the branch of medicine dealing with the use of x-rays and other types of radiant energy to diagnose and treat diseases.

**Respiratory Chain:** a series of mitochondrial proteins that work together to make energy for the body in the form of a molecule called ATP.

**Respiratory Chain Disorders:** can affect the liver to cause acute liver failure with lactic acidosis (acid build up in the blood) or more chronic liver disease with fat storage in the liver. Please see the Information for Families section on Mitochondrial Hepatopathy.

**Rickets:** A condition of infancy and childhood caused by a lack of Vitamin D and resulting in poor bone development.

**Roux-en-Y:** See Kasai Procedure.

## S

**Serum GGT:** A blood test that is used to characterize liver injury - it can be especially useful for PFIC.

**Small Intestine:** Organ where most digestion occurs. It includes the duodenum, jejunum, and ileum.

**Spinal Cord:** A tube of nervous system tissue that goes from the brain to the rest of the body.

**Spinal Tap:** A medical procedure in which fluid around the spinal cord is removed through a needle placed through the back.

**Spine:** A series of bones called "vertebrae" that make up the backbone.

**Spleen:** A large, gland-like organ located in the upper part of the abdomen on the left side of the



body. The spleen can produce oxygen-carrying red blood cells when the bone marrow is unable to do so. It also stores red blood cells for use in times of need, such as during exertion, pregnancy, severe bleeding or carbon monoxide poisoning. The spleen will enlarge when the body is diseased.

**Splenomegaly:** Enlarged spleen.

**Stones:** Small masses of tissue that form in some of the body's internal organs and can cause abdominal pain or disease.

**Stroke:** A sudden and severe attack due to a blockage or break of a blood vessel in the brain. This causes a lack of blood to flow to the brain and can lead to unconsciousness, paralysis or brain damage.

**Surgical Bypass:** A surgical procedure that bypasses part of the small intestine so that all of the bile that is made by the liver is not reabsorbed.

**Symptoms:** Any indication of disease perceived by the patient. For example, jaundice is a symptom of liver disjunction.

## T

**Thyroid:** A large gland located in the front of the neck that produces hormones vital to normal growth and metabolism.

**Tissues:** A part of an organ in the body.

**Toxic:** Poisonous.

**Toxin:** A poison that can enter the body and cause illness or disease.

**Transplant:** An organ or tissue taken from the body and implanted into another part of the same person, or into a different person.

## U

**Ultrasound Test:** An imaging test using sound waves to show the shape and size of the liver, gall bladder, bile ducts and liver blood vessels.

**Unconsciousness:** An abnormal state of lack of responsiveness to sensory stimuli, resulting from injury, illness, shock, or other bodily disorder.

**Urine:** The liquid containing water and waste products that is eliminated from the body. Samples of urine can be tested to check the levels of many chemicals and substances in the body.



**Ursodeoxycholic Acid:** A man-made bile acid used in treating liver diseases.

## V

**Varices:** Stretched veins such as those that form in the esophagus from cirrhosis.

## W

**Wax and Wane:** To come and go; "wax" means to increase and "wane" means to decrease.

**Wheezing:** Breathing with a rasp or whistling sound caused by constriction or obstruction of the throat or tubes to the lungs.

## X

**X-Ray:** An imaging test that shows pictures of bones and other structures within the human body.