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.

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

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

**Acholic**: A lack of color in the stool because of the 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 sudden onset and a short course.

**Acute Liver Failure**: A sudden serious failure of the liver to perform its normal functions.

**Alpha-1-antitrypsin**: A normal protein which is made in the liver but put out into the blood stream to balance out the activity of another protein (trypsin). When alpha-1-antitrypsin is not made correctly it can build up in liver cells and cause liver damage. 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 not to use electricity and motor vehicles.

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

**Anastomosis (AN-nah-stuh-MOH-sis)**: A surgery 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 block the effects of histamine, a normal chemical in the blood that is causes 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 nondominant) gene from both parents.

**B**

**Benign:** Not likely to become worse or cause death

**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

**Bile Acids/Bile Salts:** Chemicals that are made by the liver from cholesterol and help form bile for use in digestion to break down fats so they can be properly used by the body.

**Bile Ducts:** The passageways through which bile moves. It is the plumbing system of the liver which connect the liver to the small intestine and pancreas.

**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).
**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)
**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:** a sample of tissue from a living person

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

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

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

**Butterfly vertebrae:** A split in the 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

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

**Cholangiography** (koh-LAN-je-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:** 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:** A normal bile acid. Along with other bile acids, cholic acid is important for digestion.

**Chromosomes:** Found in all human body cells, chromosomes are the packages that contain genes

**Chronic:** Happening over 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)
 Colon: 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.

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.

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

Diagnosis: The name doctors use to describe the cause of an illness or disease.
**Diagnostic Tests:** Medical tests that help find the cause of illness, injury or disease.

**Diarrhea:** Rapid movement of stool through the intestine, leading to watery stools and poor absorption of foods vitamins

**Digestion:** The process the body uses to break down food into simple enough to be absorbed for energy, growth, and good health.

**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)

**Disabling:** decreasing the ability to function normally.

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

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

**DNA:** Deoxyribonucleic acid- this is chemical that uses a special code to make up genes. DNA is found in chromosomes.
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 biliary 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: The process of getting rid of a waste product from the body

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

F

Failure to Thrive: The term used when baby does not grow normally.

Familial: Referring to a family unit, as with diseases that likely to occur in more commonly in a given family

Fat-soluble vitamins: This term includes vitamins A, D, E and K. They need bile for absorption by the intestine. When a patient is jaundiced there may be less bile in the intestine to absorb those vitamins and jaundiced patients require higher amounts of fat soluble vitamins.

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

Fetal: A term used to describe the unborn child during pregnancy from about 8 weeks to birth.

Floppiness (Low muscle tone): having weak muscles that causes the child to have poor control moving or supporting themselves.
**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 material that forms in the gallbladder and can cause abdominal pain.


**Gene Testing:** Testing that is done on DNA samples to see if there are changes in a gene that can cause a disease.

**Genes:** Genes are made up of DNA and are a part of chromosomes. They carry all the information (instructions) to make all the building blocks to make a person’s bodily functions correctly. Genes passed on from parents to their children. Some genes determine eye or hair color. Other genes make things work in the body and so if there is something wrong with the genes, they can cause diseases.

**Genetic:** Conditions or traits that are 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:** This is a general term that means inflammation or irritation of the liver. It has many causes.

**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 disease causing condition of the liver

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. This condition may cause the skin and with whites of the eyes to look yellow and happens 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 rantidine (Zantac).

Immunosuppression: when the body’s defenses against infections are reduced usually by using medicines.

Inborn Error: A condition that comes from an error in a gene that changes how a particular chemical in the body is made or gotten rid of by the body and then leads to disease.

Infections: When a place in the body is injured or harmed by a foreign organism or toxin, like a bacteria, fungus or virus.

Inherit: getting traits from parents

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

Intestinal Mucosa: The surface lining of the intestines

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.

Jaundice: A medical word used to describe the yellowing of the whites of the eyes and skin when there are elevated bilirubin levels in the blood. See also Hyperbilirubinemia
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 hepatopancreatoduodenostomy.

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

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 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, helps 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.

Magnetic Resonance Cholangiopancreatography (MRCP): a type of picture of the bile ducts, pancreatic duct, pancreas, liver and gallbladder that uses a special machine (not x-rays).

Magnetic Resonance Imaging (MRI) Test: uses a machine that takes pictures of internal structures in the body to determine how they are functioning without using x-rays.

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

Malnutrition: Poor nourishment caused by poor diet or a medical problem 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 series of lenses that magnifies or makes larger things that are looked through it and is used to look at human tissues.

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 without using x-rays.

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

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 (chromosomes and genes) that directs the actions of the cell.
Obstruction: A blockage of a tube system in the body that prevents the flow of liquids or solids.

Off-Label Indication: A way of using a drug or medicine that is not exactly the reason it was first developed and approved by the Food and Drug Administration. 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.

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 and between the spleen and the large intestine. It helps the body break down proteins, carbohydrates and fats, and it also makes insulin.

Parenteral Nutrition: A way to provide a liquid mixture through a special tube in a large vein that gives nutrition. It is also called hyperalimentation or total parenteral nutrition (TPN).

Partial Cutaneous Biliary Diversion: A surgical procedure that carries 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: The branch of medicine that cares for children.

Peritoneum: The lining of the abdominal cavity.

Peritonitis: Infection of the peritoneum.

Peroxisome: A small specialized part of the cell that is involved in making bile acids and other activities.

PFIC (Progressive Familial Intrahepatic Cholestasis): A term to describe a family of diseases that have jaundice, itching, and poor absorption 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 decreased levels of the protein in the blood.
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. Pruritis is common in many skin disorders, but also with diseases like diabetes and liver disorders.

Puberty: The period of life when children develop adult features and their bodies become able to produce children. 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 involved in the breakdown of glucose.

Radiologist: A physician who specializes in radiology, which is the branch of medicine dealing with the use of x-rays and other types of imaging.

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.
**Serum GGT:** A blood test that is used to see what is causing 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.

**Splenomegaly:** A spleen that is larger than normal.

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

**Stroke:** A sudden and severe decrease in blood flow to an area of the brain due to a blockage or break of a blood vessel.

**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.

**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 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 situation when a person does not respond to actions that should cause them to be alert and respond.

**Urine:** The liquid containing water and waste products that is made by the kidneys and released 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 narrowing 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.