Glossary of Medical Terms – Lowe Syndrome

Acidosis – A condition of acid-base imbalance characterized by a blood pH level that is lower than normal.

AFOs – Ankle Foot Orthotics – used to control instabilities in the lower limb by maintaining proper alignment and controlling motion

Allele – One of two or more alternative forms of a gene. It is any of several forms of a gene, usually arising through mutation, that are responsible for hereditary variation.

AST/ALT – Blood test measuring liver enzymes. The concentration of these can be high in Lowe Syndrome even without liver disease.

Amino Acids – A group of 20 small molecules that link together in long chains to form proteins. Often referred to as the "building blocks" of proteins.

Amniocentesis – A prenatal test in which a small amount of amniotic fluid is removed from the sac surrounding the fetus for testing. Prenatal diagnosis method using cells in the amniotic fluid for genetic or biochemical studies.

Animal model – A laboratory animal useful for medical research because it has specific characteristics that resemble a human disease or disorder. Some animal models are natural, and some are created by scientists by transferring genes.

Antiepileptic Drugs (AEDs) – Medications used to control seizures.

Bicarbonate (CO2) – Responsible for maintaining the balance of acids and bases in your body, as in pH balance. Bicarbonate is a base and works as an acid buffer.

Buphthalmos – A disorder of infancy marked by the visible enlargement of the eyeball occurring at birth or soon after due to uncontrolled elevated pressure inside the globe.

Carrier (genetic) – A person whose genetic make up includes the gene for a specific condition, is capable of passing on the genetic mutation, but who may or may not display disease symptoms.

Cataract – Any change in or loss of the transparency or opacity of the lens inside the eye, whether it affects vision or not.

Cell free DNA testing – Prenatal maternal blood sample used to screen for the increased chance of specific chromosome problems.

Chromosome – Structures in the nucleus of a cell which are sets of linear DNA on which the genes are arranged, carrying all the instructions for a species.

Congenital – Any trait or condition that exists from birth

Cornea – The transparent structure that forms the front part off the eye and that covers the chamber in front of the iris and pupil.

Creatinine – A compound/waste product which can be measured in the blood or urine and often determines kidney function or disease.

Cryptorchidism – Undescended testes at birth.

CT Scan (sometimes referred to as a CAT scan) – computed tomography, an x-ray technique that uses a computer to construct images of the body.

CVS (Chorionic Villus Sampling) – a prenatal test that involves taking a sample of tissue from the placenta to test for chromosomal abnormalities and other genetic problems.

Cysts – Closed capsule or sac like structures typically filled with liquid, semi solid or gaseous material, the majority of them being benign.

Dialysis – A method for removing waste such as urea from the blood when the kidneys can no longer do the job. The two types of dialysis are hemodialysis and peritoneal dialysis. In hemodialysis the patient's blood is passed through a tube into a machine that filters out waste products. The cleansed blood is then returned to the body. In peritoneal dialysis, a special solution is run through a tube into the peritoneum, a thin tissue that lines the cavity of the abdomen. The body's waste products are removed through the tube.

Diuretic – Any substance/drug that increases the flow of urine

DNA (Deoxyribonucleic Acid) – The primary genetic material of all cells that provide the blueprint for all the structures and functions of a living being.

EEG (Electroencephalography) – A neurological test that measures the electric activity of the brain. It can detect abnormalities and help evaluate seizure disorders.

Endocrinologist – A physician who diagnoses and treats individuals with hormone problems.

Enzyme – A special type of protein that helps the body's chemistry work better and more quickly by speeding up a chemical reaction or causing a chemical change in another substance.

Epilepsy – A chronic medical condition defined by recurrent seizures.

Fanconi Syndrome – A condition seen in several diseases (including Lowe Syndrome) in which substances filtered by the kidneys are excreted rather than reabsorbed due to abnormal tubules.

Fracture – A break, usually in the bone.

Gene - Specific DNA sequences (allele) on a chromosome that reside in cells and mitochondria that contain the code for a specific product, such as an enzyme or structural protein.

Genetic counselor – A medical professional (not a physician) specializing in working with patients and families with genetically inherited conditions. Genetic counseling may include a discussion and analysis of a person's family tree and testing procedures.

Geneticist – A geneticist is a physician and or scientist specializing in the study of genes and in the treatment of genetic disorders.

Genome –All the genetic material of an organism.

Glaucoma – A disorder of the eye associated with elevation of the pressure within the eye. Glaucoma damages the optic nerve and, if not controlled, causes impaired vision or blindness.

Glomeruli – The part of the kidney that filters the blood.

Golgi Apparatus – A specific part of the cell that is active in the modification and transport of proteins.

G- tube (Gastrostomy Tube) – A tube, which is surgically placed directly into the stomach, through the abdominal wall to help facilitate feeding.

Hernia – The protrusion of part of an organ through an abnormal opening.

Hypermobile – Greater than normal range of movement of joints.

Hypercalcuria - Excess excretion of calcium in the urine that can lead to kidney stones and deposition of calcium in the kidney.

Hypotonia – Poor or low muscle tone, such as seen in floppy babies.

Inborn errors of metabolism - Inherited diseases resulting from alterations in genes that code for enzymes.

Keloid – A thick scar resulting from excessive growth of fibrous tissue.

Kidneys – Two organs in the back part of the abdomen that clean waste from the blood and pass them out of the body as urine. They also control the level of some chemicals in the blood, such as hydrogen, sodium, potassium, phosphate and calcium.

L- Carnitine – A small molecule protein responsible for the transport of long chain fatty acids into.

LFTs – Abbreviation for liver function tests (blood test)

Magnetic Resonance Imaging (MRI) – An imaging technique in which magnetic energy is used to examine tissues in the body, and the information is used by a computer to create an image.

Metabolite – Product of metabolism.

Mosaicism (genetic) – When a person has two or more genetically different sets of cells in his or her body. If those abnormal cells begin to outnumber the normal cells, it can lead to disease that can be traced from the cellular level. Mosaicism happens because at least one mutation occurs at some point after the zygote is created.

Mutation – A change (in the number, arrangement, or molecular sequence) in the sequence of DNA coding in a gene.

Nephrocalcinosis – Calcium deposits in kidney tissue.

Nephrolithiasis – Calcium deposits in the urine collecting system of the kidney (kidney stones).

Nephrologist – A medical doctor who sees and treats people with kidney diseases.

Neurologist – A medical doctor who sees and treats people who have problems of the nervous system, including brain, spinal cord and peripheral nerves.

NG (Nasogastric) tube – A method to facilitate feeding in which a tube is place through the mouth or nose into the stomach.

NIH (National Institute of Health) – A U.S. government supported biomedical research center made up of 27 institutes and centers in Bethesda, Maryland. They also provide research grants for investigators in other institutions. Lowe Syndrome research has taken place in the National Human Genome Research Institute.

Nystagmus – A repetitive, involuntary, rapid, rhythmical oscillation of the eyeballs, in either the horizontal, vertical, or rotatory direction.

OCRL – Oculo-Cerebro-Renal syndrome of Lowe (Lowe Syndrome), so labelled for its involvement in the eye, brain, and kidney.

Ophthalmologist – A physician in the branch of medicine specializes in the diagnosis and treatment of disorders and refractive errors of the eyes.

Orthotics –The branch of medicine that deals with the provision and use of artificial devices such as splints and braces.

Osteoporosis – Decreased density and strength of bones that can lead to increased risk of fractures.

OT (Occupational therapy) – A branch of healthcare that helps people of all ages who have physical, sensory, or cognitive problems. It can help them to improve independence in all areas of their lives.

Phosphorus – A mineral in your body which main function is formation of bones and teeth. It is also needed to make protein for growth, maintenance and repair of cells and tissue.

Prenatal – Before birth.

Prenatal Diagnosis – Diagnosis before birth. Main methods are amniocentesis, chorionic villus sampling (CVS), and maternal blood. These tests help find genetic disorders before birth.

Prognosis – What is likely to happen in the future due to a disease or state of the disease.

PT (physical therapy) – The treatment of disease, injury, or deformity by physical methods such as massage, heat treatment and exercise rather than drugs or surgery.

Puberty – The developmental process which individuals develop adult sexual characteristics and function.

Reflux (Gastroesophageal Reflux or GER) – A condition in which the sphincter muscle at the top of the stomach does not stay shut, allowing food or liquids to come back up the esophagus.

Refraction (test) – A part of a complete medical examination of the eyes to determine whether an individual has normal vision. It is also used to determine the prescription for glasses or contact lenses.

Retina – A thin layer at the back of the eyeball containing cells that are sensitive to light and that trigger nerve impulses that pass through the optic nerve to the brain, where visual images are formed.

Renal – A term that means having something to do with the kidneys

Rickets – An abnormality of calcium and phosphorus deposition in bones due to deficiencies in minerals or Vitamin D that leads to abnormalities in bone shape and strength.

Scoliosis – Curvature of the spine.

Seizures – Disturbances in brain function, manifested as episodic impairment or loss consciousness, abnormal often jerky movement, or sensory disturbances which are caused by disturbances in the electrical activity of the brain.

Strabismus – A visual defect in which the two eyes do not align on the same object at the same time. That malalignment may occur when one or both eyes deviate inward (called 'esotropia'), or one or both eyes deviate outward (called 'exotropia') or deviate vertically.

Syndrome – A collection of many features or signs associated with a single disease.

Testosterone – The sex hormone secreted by the testes, that stimulates the development of male sex organs, secondary sexual traits, and sperm.

Tubule – Part of the kidney in which reabsorption takes place.

X chromosome – is one of two sex chromosomes. Females have two (2) X chromosomes, while males have one (1) X chromosome and one (1) Y chromosome.

X linked – a trait where a gene is located on the X chromosome.

Y chromosome – a sex chromosome which is present normally only in male cells