

Loeys-Dietz Syndrome Glossary

MEDICAL SPECIALISTS

Allergist/immunologist: doctor who specializes in allergies and/or the immune system

Audiologist: healthcare professional who performs a variety of hearing exams

Cardiologist: doctor who specializes in the structure and function of the heart

Cardiothoracic Surgeon: surgeon who performs operations to repair the heart and the aorta

Dietitian: (also dietician) expert in food and nutrition who helps promote healthy eating habits, supervises food preparation and develops specific diets to meet one's special medical needs

ENT: Ear Nose and Throat Specialist (see otolaryngology)

Gastroenterologist: doctor who specializes in the digestive tract

Geneticist: doctor who diagnoses genetic conditions

Genetic Counselor: genetics expert who supports families through education, advocacy, resources and health management

Neurosurgeon: doctor who specializes in surgery of the brain and spinal cord

Nutritionist: professional who has knowledge about nutrition and diets

Occupational Therapist: professional who works with individuals with disabilities to maximize daily living skills

Ophthalmologist: doctor who specialized in diseases of the eye and ocular nerve

Optometrist: health care professional that provides ocular exams and manages certain eye diseases

Orthotist: individual who creates orthotics, or devices such as foot and back braces to support orthopedic problems

Otolaryngologist: surgeon who specializes in the ears, nose and throat regions (commonly an ENT)

Orthopedic Surgeon: surgeon who treats bone-related conditions and uses x-rays, orthotics and/or surgical intervention to follow bone abnormalities

Physician Assistant (PA): advanced practitioner, licensed to practice medicine under the supervision of a licensed physician

Physical Therapist: professional who works with individuals with disabilities to maximize movement and functional abilities

Speech Therapist/Speech-Language Pathologist: professional who works with individuals who have a variety of speech, language, or swallowing problems

Vascular Surgeon: surgeon who operates on arteries outside of the aorta (not commonly arteries of the brain)

CARDIOVASCULAR (HEART & CIRCULATORY SYSTEM)

Aneurysm: widening or dilation of the aorta or other artery



Angiotensin: substance produced by the kidney to constrict the arterioles and drive up blood pressure

Aorta: largest artery of the body that sends blood from the heart to the rest of the body

Aortic Root: base portion of the aorta as it leaves the heart

Aortic Valve: heart valve between the left ventricle and the aorta; comprised of three flaps called cusps

Arrhythmia (or Dysrhythmia): abnormal rhythm of the heart

Arterial Tortuosity: twisting or spiraled arteries (In LDS, this most often occurs in the vessels of the neck)

Arterial tree: all the arteries in the body

Arterioles: small, muscular branches of arteries

Artery: type of blood vessel that carries blood from the heart to the all the parts of the body

Atria (singular--atrium): heart's two upper chambers, right and left

Bacterial Endocarditis: infection of the heart lining or valves

Bicuspid Aortic Valve (BAV): aortic valve that only has two flaps instead of the normal three

Bradycardia: slow heart rate (less than 60 beats per minute)

Carotid Artery: major artery in the neck that carries blood from the heart to the brain

Cerebral Hemorrhage: bleeding into the tissue of the brain from a ruptured blood vessel

Cerebral Thrombosis: blood clot in the brain tissue that blocks the flow of blood

Cerebrovascular: circulatory system of the brain

Cerebrovascular Accident (CVA): stroke or other injury to the brain, which is the result of blocked blood flow

Congenital: condition existing at birth (birth defect)

Congestive Heart Failure: heart failure in which the heart is unable to circulate blood to the body

Coronary Arteries: two arteries, right and left, that rise off the aorta and curve down over the top of the heart; provide the heart muscle with oxygenated blood

Dissection: tear or rupture in the lining of the aorta or other artery that can affect blood flow to parts of the body

Heart Murmur: abnormal sound in the heart caused by floppy heart valves or holes in the heart walls

Mitral Valve: heart valve between the left atrium and ventricle

Myocardium: muscular wall of the heart; contracts to pump blood out of the heart and then relaxes as the heart refills with returning blood

Pericardium: membrane that surrounds the heart

Pericarditis: inflammation of the pericardium

Prophylactic antibiotics: antibiotics taken before dental or other invasive procedures to avoid contamination of the bloodstream

Regurgitation: leaking of blood through a floppy heart valve

Septum: muscular wall that divides the right and let sides of the heart

Supraventricular Tachycardia (SVT): rapid rhythm of the heart that arises from the electrical system of the atria

Ventricle: lower chambers of the heart; right ventricle pumps blood to the lungs while left ventricle pumps blood to the rest of the body

Ventricular Tachycardia: rapid rhythm of the heart that arises from the electrical system of the ventricles

Vertebral Artery: major artery in the neck that carries blood from the heart to the brain



HEART DEFECTS

Atrial Septal Defect (ASD): hole between the atria (upper chambers) of the heart that allows blood to pump back to the lungs instead of the rest of the body

Mitral Valve Prolapse (MVP): floppy valve between the left atrium and ventricle

Patent Ductus Arteriosus (PDA): continued opening between the aorta and pulmonary artery which causes oxygenated and un-oxygenated blood to mix May be seen at birth, but should close in infancy

Patent Foramen Ovale (PFO): opening into the right atrium that allows veins to bring blood to the heart (skipping the lungs) when the baby is in the womb It normally closes after birth, but can remain open

Ventricular Septal Defect (VSD): hole between the ventricles (lower chambers) of the heart that allows blood in the ventricles to mix and not get pumped efficiently to the rest of the body

CRANIOFACIAL (HEAD & FACE)

Bifid Uvula/Broad Uvula: split or broadness in the tissue that hangs in the back of the throat

Blue Sclera: blue tinge to the whites of the eyes

Cleft Palate: opening or gap in the roof of the mouth; can be covered by a mucous membrane called submucous cleft

Craniosynostosis: early fusion of the skull bones

Hypertelorism: widely spaced eyes

Malar Hypoplasia: flat cheek bones

Micrognathia: small chin

Retrognathia: receding chin

Palpebral fissures: slanting of the eye openings

GENETICS

Chromosome: singular strand of DNA that contains thousands of genes; humans have 46 chromosomes

Deoxyribonucleic Acid (DNA): hereditary material that is packaged to make genes that tell our body how to grow and develop

Gene: specific sequence of DNA located on a chromosome that creates a protein to perform a specific function in the body

Genotype: genetic information of a person

Phenotype: outward, observable characteristics of a person

TGFBR1: transforming growth factor beta receptor 1

TGFBR2: transforming growth factor beta receptor 2

Autosomal Dominant: inheritance pattern where one gene of a gene pair has a mutation which causes the disorder

Autosomal Recessive: inheritance pattern where both genes of gene pair requires a mutation to cause the disorder

Mutation: change in a gene that negatively impacts its structure or function



MEDICATIONS

Blood Pressure Medications: medication that reduce blood pressure and stress on aorta and other arteries

Angiotensin II Receptor Blocker (ARB): medications that block the action of a chemical angiotensin II, which normally causes blood vessels to narrow; a chemical ARB block the AT1 receptor pathway; example: Losartan, Irbesartan

Beta-blockers: medications that reduce nerve impulses to the heart and blood vessels; makes the heart beat slower and with less force, lowering blood pressure; example: Atenolol, Propanolol

Ace inhibitors (Angiotensin Coverting Enzymes): medications that prevent the formation of a chemical called angiotensin II, which normally causes blood vessels to narrow; block the AT1 and ATII receptor pathways; cause the vessels to relax and blood pressure to go down; example: Enalapril, Lisinopril

Calcium Channel Blockers: medications that keep calcium from entering the muscle cells of the heart and blood vessels; causes the blood vessels to relax and blood pressure to go down; example: Verapamil

NEUROLOGICAL

Arnold Chiari Malformation: downward displacement of a portion of the brain (cerebellum) into the spinal cord area

Dural Ectasia: bulging or widening to the sac that contains the spinal cord

Hydrocephalus: fluid collection in the brain

SKELETAL

Arachnodactyly: long, thin fingers

Cervical Spine Instability: instability in the vertebrae directly below the skull

Clubfoot/Talipes Equinovarus: congenital deformity of the foot causing the heel to point downward and the forefoot to turn inward

Congenital Hip Dysplasia: abnormally developed hip that leaves the hip joint unstable with the possibility of hip dislocation

Contracture: tightening of muscle, tendons, ligaments or skin that prevents normal movement

Halo: piece of equipment that encircles the neck, attaches to the cervical spine with metal pins, and allows for stabilization of the spine after surgery

Kyphosis: outward curvature of the spine

Lordosis: inward curvature of the spine

Osteoporosis: poor mineralization of bones leading to decreased bone mass and fragile bones

Pectus Carinatum: protruding chest wall

Pectus Excavatum: chest wall that sinks in

Scoliosis: s-like curvature of the spine

Spinal fusion: surgical procedure to stabilize vertebrae by fusing them together

Spine: back bones

Spondylolithesis: spinal condition where one vertebrae slips forward or backward in relation to the next vertebrae; symptoms can include low back pain, pain and or muscle spasms in the thighs and lower leg, muscle weakness, and or tightness in the hamstring muscle of the leg; diagnosis is made from x-ray; can be congenital or develop over time



Sternum: chest or breast bone
Vertebrae: bones of the spine
Cervical Spine: bones in the neck area
Thoracic Spine: upper spine

SKIN

Translucent skin: when veins are easily visible under skin
Hernia: protrusion of an organ or body part through a hole in skin or connective tissue
Striae: stretch marks of the skin

SPECIAL MONITORING

Holter Monitor: machine continuously records heart rhythms
Cardiac Catheterization: insertion of tubes into a blood vessel and threaded to heart to monitor blood flow
Cardiac Computed Tomography (CT), Computerized Axial Tomographic (CAT) Scan: X-ray imaging with or without contrast dye to examine internal organs, bone, soft tissue and blood vessels
Echocardiogram: ultrasound of the heart
Electrocardiogram (ECG or EKG): recording of the electrical activity of the heart over time
Electroencephalogram (EEG): recording of the electrical activity of the brain over time
Heart-Lung Machine: bypass machine; piece of equipment that oxygenates and circulates blood for the person while the heart is opened for repair
Intubation: when a tube is inserted down the throat for breathing purposes
Magnetic Resonance Angiogram (MRA): imaging with contrast that uses a magnet and radio wave pulses to produce pictures of the arteries in the body
Magnetic Resonance Imaging (MRI): imaging without contrast that uses a magnet and radio wave pulses to produce pictures of the organs and soft tissue of the body

