

# 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

