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Loeys-Dietz Syndrome Head-To-Toe

The features of Loeys-Dietz syndrome can vary widely between individuals. Each person with Loeys-Dietz syndrome will not have every feature listed in the Head-To-Toe factsheet and this list is not all-inclusive of features described in LDS. In all cases, the LDSF recommends that you consult your own physician regarding any questions about diagnosis, management or treatment.



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Loeys-Dietz syndrome (LDS): Head-To-Toe

AREA OF THE BODY	MEDICAL SPECIALTY	MEDICAL CONDITION	DEFINITION	SYMPTOMS	DIAGNOSTIC TEST	TREATMENT	FREQUENCY
Brain/Nervous system	Neurology	Seizures	A sudden convulsion	Uncontrolled shaking of extremities or body. May present as abnormal staring episodes	EEG	Medication	Rare
	Neurology	Headaches	A continuous pain in the head			Remain well hydrated. Avoid vasoconstrictor medications. Keep a headache diary to try and identify triggers that can be avoided. Consider consultation with headache clinic.	Common
	Neurosurgery	Hydrocephalus	Increased fluid collection around the brain	Symptoms in infancy: Increasing head circumference, a bulging "soft spot" on the top of the head, vomiting, sleepiness, irritability, seizures, eyes fixed downward (sunsetting of the eyes), developmental delay.	MRI or CT	If significant, may require shunt placement or other surgery	Rare
	Neurosurgery	Dural ectasia/tarlov cysts	Bulging or widening to the sac that contains the spinal cord	Usually non-symptomatic. Symptoms may include change in bowel or bladder behavior, tingling /loss of sensation/shooting pain down lower extremities	MRA, CTA or myelogram	Surgery typically not indicated as it may recur. This finding can rarely be associated with dural leaks, which cause spinal fluid leakage and may require hospitalization or surgical repair of leak. Symptoms of a leak include extreme headaches, typically impacted by change of positioning.	Common finding; rarely symptomatic
Emotional	Psychology or Psychiatry	Depression or anxiety	Short or long term feelings of excessive sadness or worry	Inability to concentrate, insomnia, loss of appetite, feelings of extreme sadness, guilt, helplessness and hopelessness, and /or excessive worry	Clinical evaluation	Many children and adults benefit from counseling as a therapeutic option. Couples and/or family therapy may be helpful. Feelings may be situational depending on age or circumstance (ie. death in family, new diagnosis, needing surgical intervention).	--
Behavioral/Cognitive	Developmental Pediatrician	ADD/ADHD or developmental delay	hyperactivity/inactivity or delay in meeting developmental milestones or educational goals		Neurobehavioral evaluation	In infancy, consider early intervention services and physical, speech, occupational therapy as needed. For older children, work with schools for appropriate school plans.	Rare
Craniofacial	Neurosurgery	Craniosynostosis	Early fusion of the skull bones	Ridging of skull bones or abnormal head shape	3D CT Scan	Surgery	Common
	ENT	Cleft palate	Opening or gap in the roof of the mouth		Physical exam	Surgery	Common
	--	Bifid uvula			Physical exam	--	Typical
Eyes	Ophthalmology	Myopia (nearsightedness)	Difficulty seeing objects at a distance	Squinting to see objects at a distance, needing to sit close to tv/ books, headaches	Ophthalmology evaluation	Glasses	Common
	Ophthalmology	Strabismus or amblyopia	Misalignment of the eyes that can cause visual deficits	"wandering eye", squinting, abnormal positioning of head for vision	Ophthalmology evaluation	Eye patching, glasses, surgery	Common
	--	Hypertelorism	Widely-spaced eyes		Physical exam	--	Typical
Teeth	Dentistry	General anomalies	Multiple cavities, discoloration and soft enamel, delayed tooth eruption or delayed tooth loss, multiple sets of teeth		Dental evaluations	Routine dental appointments and intervention as indicated by Dentist.	Common
Sinus	Allergy & Immunology	Environmental allergies	An exaggerated reaction by our immune system in response to bodily contact with foreign substances in the air	Runny nose, post nasal drip, sneezing, itchy nose or eyes, asthma, wheezing, excema	Allergy testing	Avoidance of allergens. Treat infections that may co-exist.	Common

AREA OF THE BODY	MEDICAL SPECIALTY	MEDICAL CONDITION	DEFINITION	SYMPTOMS	DIAGNOSTIC TEST	TREATMENT	FREQUENCY
Stomach or digestion	Gastroenterology	Inflammatory bowel disease	Inflammation of large or small intestines (bowel)	Abdominal pain, diarrhea, weight loss	Endoscope & biopsy	Medication, food avoidance, significant disease may require G-tube placement to help with caloric intake.	Rare
	Gastroenterology	Eosinophilic esophagitis	Inflammation of the esophagus characterized by filling of esophagus with large number of white blood cells	In adults, difficulty in swallowing solid food (feeling of food "getting stuck"), chest pain. In children, abdominal pain, nausea, vomiting, coughing, and failure to thrive.	Endoscope & biopsy	Medication, food avoidance, significant disease may require G-tube placement to help with caloric intake	Common
	Gastroenterology	Food allergies	Abnormal response to food that is triggered by a specific reaction in the immune system	Itching or tingling in mouth, difficulty swallowing, nausea, vomiting, diarrhea, hives, eczema, anaphylaxis response.	History, elimination diet, allergy testing	Food avoidance. Treat asthma or eczema that may co-exist. If exposed to allergen and non-life threatening symptoms treat with benadryl. If life-threatening reaction, Epi-Pen is used.	Common
Back / spine	Orthopedics	Cervical spine malformations or instability	Bones in the vertebrae directly below the skull may not be formed or aligned correctly causing instability around the spinal cord	Inability to hold head up or abnormal head positioning. May be an asymptomatic finding.	Flexion-extension x-rays of the neck	Depending on severity, treatment may range from avoidance of tumbling or neck manipulation to surgical intervention (cervical spine fusion)	Malformations common; instability requiring surgery is rare
	Orthopedics	Scoliosis or kyphosis	Curvature of the spine	Visible curvature of spine or asymmetry of back	Physical exam and x-ray	If significant, may require bracing or surgery.	Common
Joints	Orthopedics	Joint flexibility		Hyperextension of joints	Physical exam	Avoidance of activity that puts stress on joints or causes dislocations or subluxations. Some individuals may find relief with soft braces over knee joints. Surgical intervention for chronic subluxations/dislocations may bring short term relief, but subluxations may continue to recur over time. Occupational or physical therapy may be useful.	Typical
	Orthopedics	Contractures	Tightening of muscles, tendons, ligaments, or skin that prevents normal movement	Inability to fully extend joints	Physical exam	Occupational or physical therapy. If significant, may require surgery.	Rare
Feet	Orthopedics	Clubfoot or varus deformity of foot	Foot is turned in or rotated sharply at the ankle		Physical exam	Occupational or physical therapy, bracing, surgery	Common
	Orthopedics	Flat feet			Physical exam	Orthotics or shoes with hard arch support	Common
Breathing / lungs	Pulmonary	Asthma	Chronic inflammation of the bronchial tubes (airways) that causes swelling and narrowing of the airways.	Shortness of breath, wheezing, coughing, tightness in chest	Pulmonary & Allergy evaluation	Medications. Avoid smoking	Common
Heart	Cardiology	Congenital heart defects	Atrial septal defect, ventricular septal defect, bicuspid aortic valve, mitral valve prolapse, patent foramen ovale, patent ductus arteriosus		Echocardiogram and Cardiology evaluation	Serial imaging, surgery	Common
	Cardiology	Aortic aneurysm	Widening or dilation of the aorta		Echocardiogram, MRA, CTA, Cardiology evaluation	Use blood-pressure lowering medications to reduce blood pressure & stress on the aorta. Exercise modifications (avoidance of contact or competitive sports, avoidance of isometric exercises and avoidance of exercising to exhaustion). Many individuals require aortic-root replacement surgery.	Typical
	Cardiology	Arrhythmia	Abnormal heart rhythm	Heart beat may feel too slow, too rapid or irregular	Holter monitor, Cardiology evaluation by electrophysiology specialist	Medications or surgical intervention	Unknown
Arteries	Vascular Surgery	Aneurysms throughout the arterial tree	Widening or dilation of arteries throughout the body		CTA or MRA	Use blood-pressure lowering medications to reduce blood pressure & stress on the arteries. Exercise modifications (avoidance of contact or competitive sports, avoidance of isometric exercises and avoidance of exercising to exhaustion). Many individuals require grafting, stenting or coiling procedures for aneurysm repair	Typical
	Radiology	Arterial tortuosity	Twisting or spiraled arteries		CTA or MRA	Diagnostic clue; tortuous arteries are not predisposed to aneurysms. Surgeons should be aware of different blood vessel anatomy for surgeries. Rarely, a tortuous artery may impinge on esophagus or trachea.	Typical

AREA OF THE BODY	MEDICAL SPECIALTY	MEDICAL CONDITION	DEFINITION	SYMPTOMS	DIAGNOSTIC TEST	TREATMENT	FREQUENCY
Bone	Endocrinology	Osteopenia or osteoporosis	Poor mineralization of bones leading to decreased bone mass and fragile bones	Decreased bone density, stress fractures	Dexa Scan & Endocrinology evaluation	Delayed bone healing may mean that orthopedic surgical intervention may require longer healing time for bone surgeries. Vitamin D replacement. Bones may be impacted by gastrointestinal problems and medications used to help other LDS features.	Common
Soft tissue	General surgery	Hernia	Protrusion of an organ or body part through a hole in connective tissue	Diagnosed by physical exam or MRA/CTA (if hernia is internal location like in the diaphragm)	Physical exam or MRI & CT if internal hernia	Surgery. Hernias can recur and use of mesh may be a consideration.	Common
Chest	General surgery or Thoracic surgery	Pectus excavatum or carinatum	Chest wall that protrudes out or caves in		Physical exam, x-ray	Some individuals choose to pursue surgical correction, however, timing of this surgery is very important to consider in conjunction with skeletal growth.	Common
Skin	--	Abnormal scarring, translucent skin, easy bruising			Physical exam	Plastic surgery to correct abnormal scars may not have optimal outcome.	Common
	Dermatology	Eczema	Dry, itchy or scaly skin		Physical exam	Topical ointment or moisturizer. This may be related to allergies, thus avoidance of allergens (food/environmental) may be helpful.	Unknown
Pregnancy	Obstetrics		**Considered high risk for risk or aortic, arterial or uterine tear or rupture**		MRA or CTA & echocardiogram prior to pregnancy. Echocardiograms in each trimester	Angiotensin receptor blocker as treatment for blood pressure management is contraindicated as it is teratogenic to fetuses; beta-blockers are recommended. Create delivery plan with High Risk Obstetrician. Consider genetic counseling to review recurrence risk or genetic testing options.	--
Menstruation	Gynecology	Abnormalities of menstruation			Gynecology evaluation	Individualized treatment plan.	Unknown