EMORY UNIVERSITY SCHOOL OF MEDICINE

Department of Human Genetics

2165 N. Decatur Rd., Decatur, GA. 30033 ▪ www.genetics.emory.edu

Lysosomal Storage Diseases: Think Genetics

Is there a patient in your care with a lysosomal storage disease? Patients see an average of 6 to 13 physicians before their condition is accurately diagnosed. Lysosomal storage diseases are a family of >40 disorders caused by inherited enzyme deficiencies which cause lysosomes to become engorged. Each disease is a consequence of the type of substrate and where it accumulates. Progressive accumulation can cause irreversible damage. Use the table below as a reference guide – don't be caught missing this diagnosis! **Call the Lysosomal Storage Disease Center at Emory to learn more: (404) 778-8565 or (800) 200-1524. See website for specimen requirements.**

Condition	Common Symptoms	Specialties the Patient May See BEFORE a Diagnosis is Made	What Testing to Order
Fabry	Proteinuria, hypohidrosis, LVH, acroparesthesia, chronic fatigue, depression, chronic diarrhea, angiokeratoma, early stroke	Cardiology, Dermatology, Gastroenterology, Nephrology, Neurology, Ophthalmology, Psychiatry	• Men: Alpha-galactosidase A enzyme • Women: Alpha-galactosidase A enzyme AND sequencing of the GLA gene
Gaucher, Type I	Anemia, thrombocytopenia, fatigue, hepatosplenomegaly, bone pain, growth retardation, osteopenia/osteoporosis	Endocrinology, Hem/Onc, Orthopaedics, Rheumatology	• Glucocerebrosidase
MPS I (Hurler/Scheie)	Stiff joints, hepatosplenomegaly, coarse features, hernia, corneal clouding, chronic rhinitis, developmental delays, gibbus deformity	ER Physicians, ENT, Ophthalmology, Cardiology, Orthopaedics, Rheumatology	• Alpha-L-iduronidase • Glycosaminoglycans (GAGs) • Oligosaccharides (urine)
MPS II (Hunter)	Stiff joints, hepatosplenomegaly, coarse features, hernia, chronic rhinitis	ENT, Orthopaedics Surgeon, Pulmonology, Cardiology	• Iduronate-2-sulfatase • Glycosaminoglycans (GAGs) • Oligosaccharides (urine)
MPS VI (Maroteaux-Lamy)	Stiff joints, hepatosplenomegaly, coarse features, corneal clouding, short stature, cervical cord compression, normal intelligence	Neurosurgeon, Orthopaedics, Neurology	• Arylsulfatase B activity • Glycosaminoglycans (GAGs)
Pompe, Infantile Onset (Acid Maltase Deficiency II, GSD II)	Infantile onset cardiomegaly, hypotonia, delayed developmental milestones, macroglossia, respiratory insufficiency	Cardiology, ER Physicians, Neurology, Pulmonology	• Acid alpha-glucosidase from skin, muscle, or blood • Oligosaccharides (urine)
Pompe, Adult Onset (Acid Maltase Deficiency II, GSD II)	Progressive muscle weakness, fatigue, gait abnormality, sleep apnea	Neurology, Pulmonology, MDA Clinic	• Acid alpha-glucosidase from skin, muscle, or blood • Oligosaccharides (urine)
Tay Sachs, Infantile Onset	Hypotonia, exaggerated startle response, loss of the ability to hold the head up or sit, cherry red spot in macula	Neurology	• Beta Hexosaminidase A
Tay Sachs, Late Onset	Ataxia, tremor, psychotic episodes	Neurology, Psychiatry, Movement Disorders Clinic	• Beta Hexosaminidase A