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.

<table>
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<tr>
<th>Condition</th>
<th>Common Symptoms</th>
<th>Specialties the Patient May See BEFORE a Diagnosis is Made</th>
<th>What Testing to Order</th>
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</table>
| 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                                                                |