LYSOSOMAL STORAGE DISORDERS

COMBINING ENZYME ACTIVITY, BIOMARKER TESTING, AND MOLECULAR ANALYSIS

TO PROVIDE PHENOTYPIC AND DIAGNOSTIC INFORMATION
WHAT ARE LYSOSOMAL STORAGE DISORDERS?

Lysosomal storage disorders (LSDs) are a diverse group of inherited diseases caused by accumulation of macromolecules in the lysosome due to defects in the macromolecules’ transport mechanisms or due to defective lysosomal enzyme function. These disorders have clinically diverse phenotypes and range in severity from death during infancy to milder, later-onset variants. LSDs can be further divided into several subcategories:

- Oligosaccharidoses
- Mucolipidoses
- Mucopolysaccharidoses
- Lipidoses
- Sphingolipidoses
- Glycogen Storage Diseases

THE IMPORTANCE OF BIOCHEMICAL TESTING

Mayo Clinic’s Biochemical Genetics Laboratory performs more than 40 qualitative and quantitative laboratory tests to measure a large and growing number of analytes related to LSDs. Enzyme analysis is considered the gold standard for diagnosis of LSDs and can provide important phenotypic information on disease severity. In addition, biomarker and other analyte testing can be used for diagnosis and/or disease management. Molecular testing is often recommended as a final step in the diagnostic workup. The combination of biochemical and molecular testing is most useful to make a diagnosis of a lysosomal storage disorder.

Biochemical testing includes the following manual, automated, and chromatographic methods:

- High-Performance Liquid Chromatography (HPLC)
- Gas Chromatography–Mass Spectrometry (GC-MS)
- Tandem Mass Spectrometry (MS-MS)
WHAT TESTING SHOULD I ORDER?

More than 40 lysosomal storage disorders have been described with a wide phenotypic spectrum. Some disorders have a distinct clinical presentation, while others disorders may be difficult to differentiate based on clinical symptoms alone. As such, Mayo Medical Laboratories (MML) offers multiple testing options to suit each clinical need. Our recommended testing approach will depend on the specific clinical presentation and the disorder(s) in question.

A screening panel may be the most appropriate first test in the initial workup for some groups of disorders where the clinical features can overlap, making targeted testing time-consuming and expensive. In cases where the clinical symptoms are strongly suggestive of a specific disorder, starting with a single enzyme, analyte, or biomarker test may be the preferred approach. In some cases, molecular testing may be indicated or desired to confirm the diagnosis. MML offers mutation analysis for well-described disorders that have common pathogenic mutations and full gene sequencing for many LSDs.

FEATURED TESTS

- Lysosomal Storage Disorders Screen, Urine (Mayo ID: LYSDU)
- Oligosaccharide Screen, Urine (Mayo ID: OLIGU)
- Mucopolysaccharides (MPS) Screen, Urine (Mayo ID: MPSSC)
- Lysosomal Storage Disease Panel by Next-Generation Sequencing (Mayo ID: LSDP)
- Psychosine, Blood Spots (Mayo ID: PSY)
- Glucopsychosine, Blood Spots (Mayo ID: GPSY)

NEWBORN SCREENING

In recent years, new treatments have emerged for several LSDs, and evidence has shown that the best outcomes are achieved when treatment is initiated early in life, preferably before the onset of clinical symptoms. With disease-specific treatments now available, and the federal recommendation to screen for Mucopolysaccharidosis Type-I and Pompe disease, several states have implemented or are considering adding one or more LSDs to their state screening panels. In states where screening is performed, diagnostic follow-up testing will be needed and is available from MML in a variety of specimen types. In states where screening is not performed, MML offers newborn screening tests for LSDs intended to supplement the state-provided panel. This testing is available to any hospitals or physicians wishing to provide this screening to their patients. MML’s newborn screening tests include second-tier biomarker and molecular testing to increase the positive predictive value and reduce the cost and anxiety caused by false positive results.

CONSULT WITH MAYO CLINIC GENETIC COUNSELORS ABOUT TEST OPTIONS

Genetic counselors bring real value to proper test utilization by making sure the most appropriate test has been ordered. Genetic counselors are available to discuss diagnostic testing options and strategies.

Mayo Clinic genetic counselors are available to:

- Support the ordering process.
- Provide additional information about testing options.
- Offer results interpretation.
- Assist with case review and coordination.
TAP INTO THE EXPERTISE OF MAYO CLINIC

The Mayo Clinic Biochemical Genetics Laboratory is an interdisciplinary group of physicians, scientists, genetic counselors, and laboratory professionals that includes six laboratory directors and four genetic counselors specializing in newborn screening and diagnostic testing for inborn errors of metabolism. This team provides testing and result interpretation of the highest quality for the diagnosis and clinical care of patients with metabolic disorders. The laboratory has a long track record of innovative test improvement and test development to achieve newborn screening with the highest sensitivity and specificity, paying particular attention to reducing false positive rates. Offering one of the most comprehensive test menus, Mayo Medical Laboratories has more than 150 qualitative and quantitative genetic assays that are available for newborn screening, diagnosis, and treatment monitoring.

FOR MORE INFORMATION ABOUT LYSOSOMAL STORAGE DISORDERS, VISIT:
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