LYSOSOMAL STORAGE & PEROXISOMAL DISORDERS
ADVANCING A NEW STANDARD OF CARE FOR NEWBORN SCREENING
CHANGING RECOMMENDATIONS FOR NEWBORN SCREENING

The recommendations and legislation surrounding newborn screening are constantly in flux. New disease treatments or new laboratory technologies often prompt additional recommendations for screening. Although each state is responsible for creating and overseeing its own newborn screening program, national recommendations from the U.S. Secretary of Health and Human Services are intended to guide the development of these programs. The Recommended Universal Screening Panel (RUSP) includes 34 primary conditions and more than 25 secondary conditions. The most recent additions to the panel include disorders that are categorized as lysosomal storage and peroxisomal disorders. An increasing number of states have already passed or proposed legislation requiring screening for one or more of these disorders, although only a handful have implemented screening.

WHAT ARE LYSOSONAL STORAGE AND PEROXISOMAL DISORDERS?

Lysosomal storage disorders (LSDs) are a diverse group of inherited diseases where macromolecules accumulate across the lysosomal membrane due to defects in the macromolecules' transport mechanisms or due to defective lysosomal enzyme function. Peroxisomal disorders include disorders of peroxisomal biogenesis and single peroxisomal enzyme/transporter defects. Both types of disorders have clinically diverse phenotypes and range in severity from death during infancy to milder later onset variants. Included in this group of disorders are the following:

- Gaucher Disease
- Pompe Disease
- Krabbe Disease
- Fabry Disease
- Niemann-Pick Disease Types A & B (NPAB)
- Mucopolysaccharidosis Type 1 (MPS-I)
- X-Linked Adrenoleukodystrophy (XALD)

Mayo Medical Laboratories offers two screening assays to detect lysosomal storage and peroxisomal disorders. Both screening assays use tandem mass spectrometry to measure the activity of six enzymes each associated with a lysosomal storage disorder. One of the screening assays, LDALD, also detects four lysophosphatidylcholines, which are associated with XALD. These screening tests also include several disease-specific second-tier assays that are performed when the primary screening test yields an abnormal result. The rationale of this approach is to increase the positive predictive value of screening, thereby reducing unnecessary follow-up cost and anxiety caused by false positive results.
A NEW STANDARD OF NEONATAL CARE

With the recent updates to the RUSP and newly passed legislation in many states, gaps exist between the screening required by state law or recommended by the federal RUSP and the screening provided by state health departments. Nevertheless, with the addition of new conditions to the RUSP, health care providers may wish to meet this new standard of care as soon as possible. Mayo Medical Laboratories’ newborn screening tests are specifically designed to help a hospital supplement the screening already provided by its state’s health department. Adding one of Mayo Medical Laboratories’ screening panels ensures that newborns receive the highest quality of care within your health care organization.

The most recent additions to the panel include Pompe disease, MPS-I and XALD. In addition to these conditions, four other LSDs have been added or are being considered for newborn screening programs, including Krabbe disease, Fabry disease, Gaucher disease, and Niemann-Pick disease types A and B (NPAB). With the exception of NPAB, disease-specific therapies are now available for these disorders. Evidence has shown that best outcomes are achieved when treatment is initiated early in life, preferably before the onset of clinical symptoms.

COMPREHENSIVE FOLLOW-UP

Positive newborn screening results require prompt follow-up to confirm a diagnosis and plan a course of clinical care and treatment. The Mayo Clinic Biochemical Genetics Laboratory offers a comprehensive test menu for lysosomal storage and peroxisomal disorders. Our test menu includes traditional enzyme activity assays, novel biomarker testing, and molecular analysis, including full-gene sequencing, mutation analysis, and known mutation testing.
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 inborn errors of metabolism and 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 for inborn errors of metabolism, more than 150 qualitative and quantitative genetic assays are available for newborn screening, diagnosis, and treatment monitoring.

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