



PerkinElmer Receives FDA Clearance for Lysosomal Storage Disorder Test

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NeoLSD™ MSMS Kit is First Commercial IVD Kit to Use Mass Spectrometry to Screen for Six LSDs in Newborn Babies from a Single Dried Blood Spot Sample

WHAT: [PerkinElmer, Inc.](#), a global leader in innovating for a healthier world, today announced that it received 510(k) clearance from the FDA to commercially market its [NeoLSD™ MSMS kit](#) in the United States. The NeoLSD™ MSMS kit is the first commercial in vitro diagnostic (IVD) kit in the United States to utilize mass spectrometry instrumentation for the six most commonly screened lysosomal storage disorders (LSDs), all from a single dried blood spot sample. Mass spectrometry, with its proven technology and high sensitivity, is the platform of choice in a majority of screening labs.

LSDs are a group of nearly 50 genetically acquired diseases with an estimated combined worldwide incidence of approximately 1 in 7000-8000 live births. These disorders result from the dysfunction, deficiency or absence of a lysosomal enzyme. Affected individuals are unable to metabolize the disease-specific substrate of the deficient enzyme, which leads to its progressive accumulation in the lysosomes of tissues.

Enzyme replacement therapy or hematopoietic stem cell transplantation are available to treat some LSDs; however, early diagnosis of LSDs by clinical observation can be difficult because symptoms are absent at birth, and irreversible tissue damage (and in some cases, death) may result if an available therapy is delayed.

HOW: The NeoLSD MSMS Kit is intended for the quantitative measurement of the activity of the enzymes acid-β-glucocerebrosidase (ABG), acid-sphingomyelinase (ASM), acid-α-glucosidase (GAA), β-galactocerebrosidase (GALC), α-galactosidase A (GLA) and α-L-iduronidase (IDUA) in dried blood spots (DBS) from newborn babies. The analysis of the enzymatic activity is intended as an aid in screening newborns for the following lysosomal storage disorders (LSD), respectively; Gaucher Disease, Niemann-Pick A/B Disease, Pompe Disease, Krabbe Disease, Fabry Disease, and MPS I Disease.

Leveraging sophisticated tandem mass spectrometry (MSMS) technology, which significantly reduces sample prep and sample analysis times, laboratories can utilize the NeoLSD™ MSMS Kit to run hundreds of tests per day on a single MSMS instrument. No other available commercial offering is able to test for this number of LSDs from a single sample. Additionally, no other offering is able to achieve the same number of LSD analysis results using a single instrument.

WHY: "PerkinElmer is the only company that offers a comprehensive IVD solution for analyzing these six LSDs from a single blood spot sample," said Naren Bhat, General Manager, Mass Spectrometry, PerkinElmer. "We continue to innovate and develop screenings for an increasing number of LSDs to meet the evolving needs of our customers."

MORE: PerkinElmer offers a global diagnostics portfolio focused on: reproductive health, infectious disease screening and genomics offerings for oncology and other molecular tests through its wide range of instruments, reagents, assay platforms and software offerings.

ABOUT PERKINELMER:

PerkinElmer, Inc. is a global leader committed to innovating for a healthier world. Our dedicated team of about 11,000 employees worldwide is passionate about providing customers with an unmatched experience as they help solve critical issues especially impacting the diagnostics, discovery and analytical solutions markets. Our innovative detection, imaging, informatics and service capabilities, combined with deep market knowledge and expertise, help customers gain earlier and more accurate insights to improve lives and the world around us. The Company reported revenue of approximately \$2.3 billion in 2017, serves customers in more than 150 countries, and is a component of the S&P 500 Index. Additional information is available through 1-877-PKI-NYSE, or at www.perkinelmer.com

Media Contact:

Brian Willinsky
brian.willinsky@perkinelmer.com
781-663-5728