PerkinElmer Expands Screening Services to Detect Lysosomal Storage Disorders in Newborns

WALTHAM, Mass.--(BUSINESS WIRE)-- PerkinElmer, Inc., a global leader focused on improving the health and safety of people and the environment, announced today that its newborn screening laboratory service, PerkinElmer Genetics, has launched a new panel to screen for six Lysosomal Storage Disorders (LSDs) as an addition to its current newborn testing and diagnostics portfolio. The LSD Screening Panel tests for Krabbe disease, Gaucher's disease, Niemann-Pick disease (Type A and Type B), Pompe disease, Fabry disease and MPS I.

"The new LSD panel is the latest addition to our diagnostic and screening services portfolio of more than 50 metabolic disorders. It enhances the ability to detect six additional genetic disorders within 72 hours of specimen receipt, enabling earlier clinical intervention for better neonatal health," said Jim Corbett, president, Diagnostics, PerkinElmer. "While the primary goal is to provide information that improves a newborn's quality of life and a definitive diagnosis for parents, such tests also have the potential to lower palliative healthcare costs for families and insurers, due to early diagnosis."

Each LSD results from different genetic mutations that translate into a deficiency in enzyme activity. However, they all share a common biochemical characteristic — all lysosomal disorders originate from an abnormal accumulation of substances inside the lysosome. The lysosome is responsible for converting cell waste into reusable matter for cell function. Enzymes catalyze the breakdown of this unwanted material, but in individuals with an LSD, the necessary enzyme is missing or exists in an insufficient amount. This causes the cell waste to accumulate in excess within the cell. Symptoms of LSDs can include muscle damage, respiratory difficulties, bone abnormalities, joint stiffness, burning sensations, enlarged liver or spleen, seizures and loss of learned skills. When left untreated, severe deficiencies may result in death.

The collective group of approximately 50 known LSDs has an incidence rate between 1 out of 5,000 and 1 out of 10,000. Although there is no known cure for LSDs, early detection allows doctors to plan for treatment which involves addressing symptoms that may occur between infancy and adulthood. Emerging therapies for LSDs include enzyme replacement therapy (ERT) and hematopoietic stem cell transplantation (HSCT) using bone marrow or umbilical cord blood as a source of healthy stem cells.

LSD screening is available as stand-alone testing in the **Lysosomal Storage Disorders Screening Packet (LSD Only)** or included with the **StepOne®/ LSD Newborn Screening Packet**.

Services offered by PerkinElmer Genetics are designed to detect treatable diseases as early as possible before irreparable damage to health occurs. PerkinElmer Genetics provides high quality newborn screening services to save and improve the quality of children's lives through early detection and intervention. PerkinElmer Genetics has established and verified its tests' accuracy and precision as required under CLIA '88. For more information about PerkinElmer Genetics, please visit: www.perkinelmergenetics.com.

About PerkinElmer, Inc.

PerkinElmer, Inc. is a global leader focused on improving the health and safety of people and the environment. The Company reported revenue of approximately $1.7 billion in 2010, has about 6,200 employees serving customers in more than 150 countries, and is a component of the S&P 500 Index. Additional information is available through 1‐877‐PKINYSE, or at www.perkinelmer.com.

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