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PerkinElmer Launches CE-IVD Assay Kit to Screen Newborns Affected by Duchenne Muscular Dystrophy

First Commercially Available Automated Immunoassay for Measuring Creatine Kinase Muscle Isozyme (CK-MM) in Dried Blood Spots

WHAT: PerkinElmer, Inc., a global leader committed to innovating for a healthier world, today announced the launch of the first commercially available in-vitro diagnostic (IVD) kit for newborn screening of Duchenne muscular dystrophy (DMD),* a progressive, X-chromosome linked neuromuscular disorder with a worldwide average incidence rate of approximately 1 in 5,000 male live births.

With DMD, mutations in the dystrophin gene lead to the absence of functional dystrophin protein in the muscles. Dystrophin supports the mechanical strength of muscle fibers, and its absence leads to progressive muscle weakness and wasting-- eventually resulting in premature death due to respiratory problems and cardiac failure. The progressive breakdown process of skeletal muscle cells releases intracellular creatine kinase (mostly the muscle specific isoform CK-MM) into the circulation, thereby allowing the level of this biomarker in the blood to be used for the early detection of the disorder.

Recent therapeutic developments for DMD, along with new evidence of the importance of early intervention with therapy (such as corticosteroid treatment), have underscored the need for newborn screening of DMD.

PerkinElmer's GSP® Neonatal Creatine Kinase –MM (CK-MM) kit is an immunoassay for measuring CK-MM in newborn babies' dried blood spot samples. The assay measures CK-MM concentration instead of enzyme activity and therefore is not susceptible to interference from inactivation during the sampling workflow. As the assay is specific to detect the muscle specific isoform, it improves the screening process to find the cases affected by DMD. The kit is available on PerkinElmer's GSP® instrument, a fully automated, high throughput biochemical analyzer that is intended for testing neonatal dried blood spot samples to detect potentially life-threatening conditions in newborn babies and enables highly efficient screening workflow.

"PPMD is committed to paving a path forward for newborn screening for Duchenne in the United States," said Pat Furlong, Founding President & CEO, Parent Project Muscular Dystrophy (PPMD), the largest and most comprehensive nonprofit organization in the U.S. focused on ending Duchenne. "We are pleased to be working closely with PerkinElmer to further effective testing methods and efficiencies that we believe will ultimately eliminate unnecessary diagnostic odysseys and delays for families with Duchenne. This technology moves us closer to our goal of providing families and providers options for early interventions and eventually treatments."

WHY: Previously, the only available alternative for biochemical assessment of CK levels in dried blood spot samples for newborn screening consisted of non-standardized lab-developed tests based on CK enzyme activity measurement.

"Using CK-MM as a viable marker for Duchenne can lead to advancements in research and potential treatment, providing children opportunities for improved health," said Linh Hoang, Vice President, Neonatal Screening, PerkinElmer. "Our introduction of the first wide scale IVD assay for routine diagnostic testing for Duchenne can dramatically improve productivity in newborn screening processes for lab professionals. They can expand their menus to include DMD screening efficiently on the GSP."

Dr. Stuart J. Moat, FRCPath, Consultant, Clinical Biochemist and Director for the Wales Newborn Screening Laboratory at the University Hospital of Wales published a study earlier this year in *Clinical Chemistry* revealing the potential of PerkinElmer's CK-MM kit in identifying Duchenne in newborn babies.

"We found that CK-MM can be reliably quantified in blood spots and believe that developing this CK-MM assay on a commercial immunoassay analyzer would enable standardized, high-throughput screening for DMD," said Dr. Moat.

MORE: As the global leader in newborn screening, PerkinElmer currently serves customers in more than 100 countries worldwide. Its customers have screened more than 600 million babies throughout the world for life-threatening diseases: screening 39 million babies annually around the world, helping to save more than 70 babies per day (one baby every 20 minutes).

PerkinElmer has also collaborated with local parties to set up a pilot program for Duchenne screening at Zhejiang Children's hospital in Hangzhou, China where around 42,000 male infants were screened for DMD. A pilot study is being initiated to evaluate the potential applicability for the U.S.

For additional information on PerkinElmer's GSP Neonatal Creatine Kinase – MM kit, please click here.

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

**PerkinElmer's CK-MM kit is currently available for research use only in the U.S. and China. It is not for use in diagnostic procedures.*

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