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New Study Published in *Clinical Chemistry* Evaluates Biomarker for Duchenne Muscular Dystrophy Screening in Newborn Babies

PerkinElmer, Inc. Provides Research-Use-Only Automated Immunoassay to Detect Creatine Kinase Muscle Isozyme in Dried Blood Spots

WHAT: A new study published in *Clinical Chemistry* has revealed a potential automated assay for identifying Duchenne muscular dystrophy (Duchenne) in newborn babies. Duchenne is a progressive, X-chromosome linked neuromuscular disorder with a worldwide average incidence rate of approximately 1 in 5,000 babies.

With Duchenne, the absence of functional dystrophin protein in the muscles leads to progressive muscle weakness and wasting, which eventually leads to premature death. The progressive breakdown of skeletal muscle cells in babies with Duchenne releases intracellular creatine kinase muscle isozyme (CK-MM) into the circulation, thereby potentially allowing the marker to be used to screen for the disorder.

The blood spot CK enzyme assays used in previous newborn screening programs for Duchenne did not specifically focus on the CK-MM protein, which is more stable than enzyme activity. Whereas prior studies measured non-specifically all forms of CK (also from other sources), this study examined a subform of CK-MM specifically found in muscle.

WHY: "The study analyzed the performance of a prototype automated immunoassay for measuring CK-MM from dried blood spots that was developed by PerkinElmer," said Dr. Stuart J. Moat, FRCPath, Consultant, Clinical Biochemist and Director for the Wales Newborn Screening Laboratory at the University Hospital of Wales. "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 could enable standardized, high-throughput newborn blood spot screening for Duchenne. The Cardiff University and Cardiff and Vale Universal Health Board Clinical Innovation Partnership have been integral to analyzing this automated method that was developed by PerkinElmer."

"The successful outcome of this study is an example of the impact that can be achieved through collaboration between academic, clinical and industry colleagues in developing improvements in human healthcare," said Professor Ian Weeks, D.Sc., Dean of Clinical Innovation and Head of the University's School of Medicine.

HOW: Based on the prior work of Dr. Moat and Professor Weeks in developing a chemiluminescent assay, PerkinElmer developed a fluorescence-based in-vitro diagnostic (IVD) kit for Duchenne screening (currently available for research use only outside of the U.S.). Using PerkinElmer's Genetic Screening Processor® (GSP) instrument, a fully automated, high throughput analyzer that tests neonatal dried blood spot samples, the study team evaluated the analytical performance of the PerkinElmer-developed prototype automated immunoassay for measuring CK-MM.

"PPMD is committed to paving a path forward for newborn screening for Duchenne in the United States. We are working closely with PerkinElmer to further effective testing methods that we believe will lead to advancements in research for early interventions and eventually treatments," said Pat Furlong, President & CEO, Parent Project Muscular Dystrophy (PPMD), the largest and most comprehensive nonprofit organization in the U.S. focused on ending Duchenne.

"At Muscular Dystrophy UK we regularly hear from families who tell us that an earlier diagnosis would help them to plan and make adjustments in the knowledge of their child's condition. Therefore, developing a diagnostic test which will enable early detection is extremely important," said Nic Bungay, Director of Campaigns, Care and Information at Muscular Dystrophy UK. "Muscular Dystrophy UK has been working with scientists such as Dr. Stuart Moat to help pave the way to newborn screening for Duchenne, and we are committed to continuing to do so. We are also working with healthcare professionals and health commissioners here in the UK to improve the emotional support available to recently diagnosed families. Any screening program needs to be accompanied by psychological support from experts in Duchenne."

MORE: PerkinElmer also collaborated with local parties to set up a pilot program in China. In Wisconsin a pilot study is being initiated to evaluate the potential applicability for the U.S.

"As the global leader in newborn screening, we are pleased to collaborate on innovative research related to helping advance the processes for screening for rare disorders such as Duchenne," said Linh Hoang, Vice President, Neonatal Screening, PerkinElmer.

"Using CK-MM as a viable marker for this very harmful disorder impacting newborn babies around the world is another step forward in giving children with this condition a better chance at improving their health."

ABOUT PERKINELMER:

PerkinElmer, Inc. is a global leader committed to innovating for a healthier world. The Company reported revenue of approximately $2.1 billion in 2016, has approximately 9,000 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-PKI-NYSE or at www.perkinelmer.com.

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