



PerkinElmer Launches First FDA-Approved Assay Kit to Screen for Duchenne Muscular Dystrophy in Newborns

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Available on PerkinElmer's automated GSP[®] instrument, kit supports early diagnosis by measuring CK-MM concentration instead of CK enzyme activity

WALTHAM, Mass.--(BUSINESS WIRE)--Dec. 13, 2019-- [PerkinElmer, Inc.](#), (NYSE: PKI), a global leader committed to innovating for a healthier world, today announced that its [GSP[®] Neonatal Creatine Kinase –MM \(CK-MM\) kit](#) has received U.S. Food & Drug Administration (FDA) approval. This solution is the first commercially available assay for screening newborns affected by Duchenne muscular dystrophy (DMD).

DMD is an X-linked recessive disease and the most prevalent type of muscular dystrophy, affecting approximately 1 in 5,000 live male births. The disorder is caused by mutations in the dystrophin gene, which supports the mechanical strength of muscle fibers. Without dystrophin, a patient's muscles progressively weaken and deteriorate, ultimately resulting in premature death from poor respiratory function and cardiac failure.

Although there is no known cure, recent studies demonstrate the importance of early intervention and treatment. In particular, steroid treatments like [corticosteroids](#) have been proven to support muscle repair and reduce detrimental side effects in infants, underscoring the need for DMD testing at the earliest point in life.¹ PerkinElmer's kit is specifically designed for screening newborn babies by measuring CK-MM—the predominant isoform in skeletal muscle cells and most specific to skeletal muscle damage—in dried blood spot samples. CK-MM levels are typically elevated in DMD patients, as the degeneration of skeletal muscle cells causes CK to release into the bloodstream.

"It is a cost-effective way to screen for DMD with a two-tier testing approach; the first tier CK-MM assay allows identification of infants with excess muscle damage at birth, and the second tier DMD gene analysis would link the muscle damage to the genetic defect," said Mei Baker, MD, FACMG, professor in the Department of Pediatrics and Co-Director in the Newborn Screening Laboratory at the University of Wisconsin School of Medicine and Public Health.

"Screening newborns not only prevents DMD patients and their families from an unnecessary diagnostic odyssey, but also ensures timely treatment for a disease that could otherwise go undetected for years," said Linh Hoang, M.D., Ph.D., Vice President, Reproductive Health, PerkinElmer. "By measuring the muscle-specific isoform, rather than total CK activity, PerkinElmer's assay enables clinicians to identify babies with this condition faster and, most importantly, give them a better chance at improved health outcomes."

For more information on PerkinElmer's CK-MM assay and other newborn screening solutions, please visit: newbornscreening.perkinelmer.com.

About PerkinElmer

PerkinElmer enables scientists, researchers and clinicians to address their most critical challenges across science and healthcare. With a mission focused on innovating for a healthier world, we deliver unique solutions to serve the diagnostics, life sciences, food and applied markets. We strategically partner with customers to enable earlier and more accurate insights supported by deep market knowledge and technical expertise. Our dedicated team of about 13,000 employees worldwide is passionate about helping customers work to create healthier families, improve the quality of life, and sustain the wellbeing and longevity of people globally. The Company reported revenue of approximately \$2.8 billion in 2018, 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.

¹ Connolly AM, Zaidman CM, Golumbek PT, Craddock MM, Flanigan KM, Kuntz NL, Finkel RS, McDonald CM, Iannaccone ST, Anand P, Siener CA, Florence JM, Lowes LP, Alfano LN, Johnson LB, Nicorici A, Nelson LL, Mendell JR. (2019). Twice weekly Glucocorticosteroids in Infants and Young boys with Duchenne Muscular Dystrophy: Steroids in young boys with DMD. *Muscle & Nerve*. 10.1002/mus.26441.

² Moat SJ, Korpimäki T, Furu P, Hakala H, Polari H, Meriö L, Mäkinen P, Weeks I. Characterization of a blood spot creatine kinase skeletal muscle isoform immunoassay for high-throughput newborn screening of Duchenne muscular dystrophy. *Clin Chem* 2017;63:908-14.

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