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New test for screening of Duchenne muscular dystrophy in newborn babies

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Researchers at Cardiff University and Cardiff and Vale University Health Board have developed a more reliable method of screening for Duchenne muscular dystrophy (DMD) in newborn babies.

In collaboration with biotechnology company PerkinElmer, they have developed a diagnostic kit that can accurately screen for the disorder by analysing neonatal dried blood spots.

Professor Ian Weeks, Dean of Clinical Innovation and Head of Cardiff University's School of Medicine, said: "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."

The new method screens for DMD by detecting an enzyme that is released into the blood when muscle fibres are damaged from the condition. Previous tests of this type were less accurate as they also detected the activity of other forms of this enzyme, two of which are also produced in high levels due to muscle trauma-related injuries, which can lead to false results during screening.

Dr Stuart J. Moat, Consultant Clinical Biochemist and Director of the Wales Newborn Screening Laboratory at the University Hospital of Wales, said: "We found that CK-MM can be reliably quantified in blood spots and believe that developing this CK-MM assay on a commercial immunoassay analyser would enable standardized, high-throughput

screening for DMD."

DMD is the most fatal common genetic disorder diagnosed in childhood. The disorder gradually causes muscles to weaken, leading to an increasing level of disability and eventually premature death. DMD almost always affects boys, with around 100 boys born in the UK with the condition each year, and about 2,500 living with the condition in the UK at any one time.

The new screening method originated from research by Professor Ian Weeks from Cardiff University and Dr Stuart Moat of Cardiff and Vale University Health Board. When PerkinElmer joined the collaboration, the research was successfully adapted to an existing PerkinElmer analyser, allowing it to be translated into a routine test that could be used globally.

Linh Hoang, Vice President, Neonatal Screening, PerkinElmer, added: "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.

"This is another step forward in giving children with this condition a better chance at improving their health."

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 US.

Pat Furlong, President and CEO, Parent Project Muscular Dystrophy (PPMD), the largest and most comprehensive non-profit organization in the US focused on ending Duchenne, said: "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."

Nic Bungay, Director of Campaigns, Care and Information at Muscular Dystrophy UK, added: "This new test moves us closer to a definitive newborn screening test for Duchenne, which will give families more time to plan for the future. We are hopeful that the rapid improvements in testing will allow the rollout of a national newborn screening programme in the coming years, which will allow for treatments to be delivered to the very young at the earliest possible stage."

Jeanette George, whose son Alex was diagnosed with Duchenne muscular dystrophy through the screening programme in Wales, said: "Having the choice to screen Alex was a positive thing for us. Knowing Alex has Duchenne has allowed us to plan ahead and to manage his symptoms. Alex gets assessed every six months, so any change in his wellbeing will be picked up immediately. I have also been given the opportunity to make the decision to change the direction of my career and to spend more time at home with my son."

The new research 'Characterization of a Blood Spot Creatine Kinase Skeletal Muscle Isoform Immunoassay for High-Throughput Newborn Screening of Duchenne Muscular Dystrophy' is published in the journal *Clinical Chemistry*.

More information: Stuart J. Moat et al. Characterization of a Blood Spot Creatine Kinase Skeletal Muscle Isoform Immunoassay for High-Throughput Newborn Screening of Duchenne Muscular Dystrophy, *Clinical Chemistry* (2017). DOI: 10.1373/clinchem.2016.268425

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