



PerkinElmer Joins European Alliance for Newborn Screening in Spinal Muscular Atrophy

January 26, 2021

Alliance aims to make SMA testing mandatory in European newborn screening programs by 2025

WHAT: [PerkinElmer, Inc.](#), a global leader committed to innovating for a healthier world, today announced that the Company has joined the [European Alliance for Newborn Screening in Spinal Muscular Atrophy](#), also known as the SMA NBS Alliance. Founded by [SMA Europe](#) – an umbrella organization that brings together 21 patient and research organizations focused on spinal muscular atrophy (SMA) from 20 countries across Europe – the SMA NBS Alliance has two primary objectives: to decrease the time it takes for a child born with SMA to be diagnosed through newborn screening (NBS), and to assist patient advocacy groups in their efforts to accelerate the identification of such children, given that early diagnosis and treatment leads to better outcomes.

[SMA](#) is a rare, progressive, neuromuscular disease that leads to immobility and results in a short life expectancy for many children diagnosed with the condition. NBS offers healthcare providers an opportunity to diagnose and treat children with SMA before motor neuron destruction occurs, giving children affected by this condition the best possible quality of life.

HOW: The SMA NBS Alliance demands that by 2025, NBS programs in Europe include a test for SMA for all newborn children. Working toward this goal, Alliance members take part in a variety of activities and are responsible for organizing events in their respective countries to request that health regulators evaluate adding SMA to newborn screening panels.

The founding members of the SMA NBS Alliance include the national SMA patient organizations that are part of SMA Europe, EURORDIS – Rare Diseases Europe, the European Alliance of Neuromuscular Disorders Associations, TREAT-NMD and several global pharmaceutical companies.

WHY: The incidence of SMA is approximately 1 in 10,000 live births and it is one of the leading genetic causes of infant death worldwide. Diagnosing SMA quickly is crucial to stopping progression of the disease, which robs infants of valuable motor neurons that allow them to walk, sit and even breathe. Early detection and diagnosis may mean the difference between life and death for an infant with a severe form of SMA.

MORE: PerkinElmer is a global market leader in neonatal screening, currently serving customers in more than 100 countries. The Company's comprehensive NBS portfolio includes tests for more than 50 conditions known to have positive outcomes when detection and diagnosis occur early with newborn screening.

In September 2020, PerkinElmer received [CE-IVD approval](#) for its [FONIS™ System](#) a RT-PCR newborn screening assay enabling simultaneous testing for SMA, severe combined immunodeficiency (SCID) and X-linked agammaglobulinemia (XLA). To learn more about all of PerkinElmer's newborn screening offerings, please visit [newbornscreening.perkinelmer.com](#).

About SMA Europe

SMA Europe is a non-profit umbrella organization which consists of 21 SMA patient and research organizations from 20 countries across Europe. SMA Europe aims to improve the quality of life of people who live with SMA, to accelerate translational research pathways in SMA, to bring effective therapies to patients in a timely and sustainable way, to encourage optimal patient care as early detection of the disease.

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 14,000 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 \$2.9 billion in 2019, serves customers in 190 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](#)

###

Media Relations:

Brianna Quinn

(781) 663-5728

brianna.quinn@perkinelmer.com