Orchard Therapeutics Celebrates Addition of Metachromatic Leukodystrophy to the U.S. Recommended Uniform Screening Panel

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Other community-led efforts to enable newborn screening for MLD advancing globally

TOKYO, LONDON and BOSTON, Dec. 16, 2025 (GLOBE NEWSWIRE) -- Orchard Therapeutics, a Kyowa Kirin company, today celebrates the addition of metachromatic leukodystrophy (MLD) to the U.S. Recommended Uniform Screening Panel (RUSP), a national guideline comprising a list of medical conditions for which the federal government recommends all newborns in the U.S. are screened for at birth. States use the RUSP to inform decisions about which conditions to include in their respective newborn screening (NBS) panels.

NBS is widely considered one of the most successful public health programs worldwide. In the U.S., approximately 1 in 500 newborns have a condition that can be diagnosed through NBS, and more than 8,000 infants annually have the potential to receive life-saving treatment due to this public health program.[i] Once a new condition is added to the RUSP, adoption and implementation is then carried out at the state level. As of today, 14 states —representing more than 50 percent of total U.S. births—have RUSP alignment legislation intended to expedite the process of adding newly approved conditions to their respective state NBS panels.

“Newborn screening will prove critical as it is the only practical means of diagnosing patients prior to the onset of symptoms, which is key to achieving optimal and equitable outcomes for children in the U.S. and their families dealing with this rapidly progressive, irreversible and ultimately fatal disease,” said Bobby Gaspar, M.D., Ph.D., chief executive officer of Orchard Therapeutics. “We commend the U.S. Health and Human Services leadership for recognizing the immense and urgent medical need to screen for MLD at birth.”

Dr. Gaspar added, “While the addition of MLD to the RUSP is a monumental step toward enabling newborn screening in the U.S., implementation now must happen at the state-level, a historically multi-year process inhibited by the lack of adequate federal and state funding. Orchard Therapeutics will continue to support community-led efforts aimed at advancing newborn screening for MLD, and we recognize the valuable contributions of researchers, physicians, patient advocates and families to help ensure the implementation of newborn screening keeps pace with biomedical innovation. This is why we will continue to apply our expertise and infrastructure to help enhance and modernize newborn screening programs in the U.S. and beyond.”

MLD is an ultra-rare and severely life-limiting neurometabolic disease that affects approximately one in 100,000 live births. It is caused by an error in the gene responsible for encoding the enzyme arylsulfatase A (ARSA) leading to neurological damage and developmental regression. In the most severe form of MLD, babies initially develop normally but in late infancy start to rapidly lose the ability to walk, talk and interact with the world around them. These children eventually deteriorate into a state which may require 24-hour intensive care, and the majority pass away within five years of symptom onset, creating an enormous emotional and financial burden on the family, care partners and healthcare systems.

“Newborn screening for MLD is so important because if a child is not diagnosed prior to the onset of symptoms, they may become ineligible for treatment, as was the case for my middle daughter Livvy, who is currently in hospice dealing with the later stages of this cruel disease,” said Kendra Riley, a Phoenix-area mother and MLD advocate. As a result of Livvy’s diagnosis, Riley’s youngest daughter, Keira, was diagnosed with MLD pre-symptomatically through sibling testing and was able to seek timely medical intervention as a result.

Riley added, “Our family’s story is far too common today, but as newborn screening for MLD becomes more widely available, future generations could be spared from having to watch one child slip away in order to save another. The combination of newborn screening to diagnose MLD at birth and treatment availability changes the trajectory of this disease for children and families.”

Last year, the U.S. Food and Drug Administration (FDA) approved the first and only therapy for eligible children with pre-symptomatic late infantile (PSLI), pre-symptomatic early juvenile (PSEJ) or early symptomatic early juvenile (ESEJ)—collectively referred to as early-onset—MLD which is marketed by Orchard Therapeutics. The same treatment was approved by the European Commission (EC) in 2020 and the UK Medicines and Healthcare products Regulatory Agency (MHRA) in 2021.

“As with many rare, life-threatening diseases, early detection and diagnosis is key to ensuring the best possible outcomes for patients,” said Barbara Burton, M.D., attending physician, genetics, genomics and metabolism at the Edwards Family Division of Genetics and Rare Diseases at the Ann &

Robert H. Lurie Children's Hospital of Chicago. "I have seen first-hand the disparity of outcomes for treated versus untreated children, and I believe we are obligated to provide those with MLD the best opportunity for a meaningful life, which is only possible with universal newborn screening."

"Having been passionately involved in the advancement of newborn screening for many severe genetic diseases, I can confidently say few conditions so obviously meet widely accepted inclusion criteria in national programs than MLD," said Michael Gelb, Ph.D., Boris and Barbara L. Weinstein endowed chair in the Departments of Chemistry and Biochemistry at the University of Washington, and whose laboratory developed the assay that enabled screening for MLD to be seamlessly implemented as part of the current panel of tests using dried blood spots (DBS). The original assay has been optimized over the past few years due to efforts of a worldwide MLD newborn screening alliance and has been shown to perform with reproducibly accurate results.

Dr. Gelb added, "The coordination to formally submit the nomination for MLD newborn screening is a prime example of how community-led efforts should be run in collaboration with industry to advance shared goals. I'm immensely proud to have played a role in making this happen and hope the U.S. is a bellwether for many other countries to add MLD to their national NBS programs."

Advancing newborn screening efforts globally

In addition, community-led efforts to enable NBS for MLD continue to advance around the world. In the U.S., Illinois, Maryland, Minnesota, Pennsylvania, and Utah have added MLD to their respective state panels. Moreover, a statewide pilot study has commenced in New York and additional states' advisory committees are currently reviewing MLD, marking significant progress which is now expected to be accelerated following the addition of MLD to the RUSP.

In Europe, following the implementation of MLD to the Norwegian NBS program in January, Sweden has published a national recommendation to include MLD in its screening program, reflecting growing international recognition of the strong evidence to support NBS for MLD. Additionally, several regional resolutions have been made in Italy, the Haute Autorité de Santé (HAS) in France has announced a formal evidence review to include MLD with a recommendation due by the end of 2026, and the Federal Joint Committee (G-BA) in Germany is due to conclude its ongoing assessment to inform potential nationwide implementation in late 2027. Meanwhile the national MLD NBS pilot in Austria continues.

About MLD

MLD is a rare and life-threatening inherited disease of the body's metabolic system estimated to occur in approximately one in every 100,000 live births based on existing literature. MLD is caused by a mutation in the *arylsulfatase-A* (*ARSA*) gene that results in the accumulation of sulfatides in the brain and other areas of the body, including the liver, gallbladder, kidneys, and/or spleen. Over time, the nervous system is damaged, leading to neurological problems such as motor, behavioral and cognitive regression, severe spasticity, and seizures. Patients with MLD gradually lose the ability to move, talk, swallow, eat and see. In its late infantile form, mortality at five or ten years from onset is estimated at 75 percent and 100 percent, respectively.[ii]

About Orchard Therapeutics

Orchard Therapeutics, a Kyowa Kirin company, is a global gene therapy leader focused on ending the devastation caused by genetic and other severe diseases by discovering, developing, and commercializing new treatments that tap into the curative potential of hematopoietic stem cell (HSC) gene therapy. In this approach, a patient's own blood stem cells are genetically modified outside of the body and then reinserted, with the goal of correcting the underlying cause of disease with a single treatment.

Founded in 2015, Orchard's roots go back to some of the first research and clinical developments involving HSC gene therapy. Our team has played a central role in the evolution of this technology from a promising scientific idea to a potentially life-transforming reality. Today, Orchard is advancing a pipeline of HSC gene therapies designed to address serious diseases where the burden is immense for patients, families and society and current treatment options are limited or do not exist.

For more information, please visit www.orchard-tx.com.

About Kyowa Kirin

Kyowa Kirin aims to discover and deliver novel medicines and treatments with life-changing value. As a Japan-based Global Specialty Pharmaceutical Company, we have invested in drug discovery and biotechnology innovation for more than 70 years and are currently working to engineer the next generation of antibodies and cell and gene therapies with the potential to help patients with high unmet medical needs, such as bone & mineral, intractable hematological diseases/hemato oncology, and rare diseases. A shared commitment to our values, to sustainable growth, and to making people smile unites us across the globe. You can learn more about the business of Kyowa Kirin at www.kyowakirin.com.

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[i] *Gaviglio et. al. Infants with Congenital Diseases Identified through Newborn Screening—United States, 2018–2020. International Journal of Neonatal Screening. 2023, 9(2), 23;* https://doi.org/10.3390/ijns9020023

[ii] *Mahmood et al. Metachromatic Leukodystrophy: A Case of Triplets with the Late Infantile Variant and a Systematic Review of the Literature. Journal of Child Neurology 2010, DOI:* http://doi.org/10.1177/0883073809341669

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