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MED13L Foundation Announces Global MED13L Day and Launches $1 million Fundraising Campaign

Entrepreneurial patient advocacy group for rare disease makes strides towards CURE MED13L

Barrington, NJ: CURE MED13L, an initiative of the MED13L Foundation, today announced May 13th as World MED13L Awareness Day and launched a fundraising campaign to raise $1 million towards the development of therapies for the single-gene disorder. MED13L is a recently identified single-gene disorder that is a leading cause of autism and intellectual disability. Other symptoms of MED13L include seizures, severe impairment of speech, motor developmental delay with poor muscle tone and ataxic-like movement, cardiac abnormalities and possible increased risk of childhood leukemia. Kelly Sexton, the mother of two MED13L children and the founder of the MED13L Foundation, notes that "MED13L has a devastating impact on families and the communities around them. CURE MED13L has suddenly created hope for interventions that can radically change the course of all our lives."

Nick Seaver, a MED13L parent, biotech investor and founder of the CURE MED13L initiative, says "we are pursuing the repurposing of existing drugs in the short term. We are also pursuing the longer-term goal of advanced gene-based therapies. The work we are doing has application to other rare disease communities beyond MED13L. As we've built momentum, we've been able to attract the interest of leading researchers in these areas and have put in place a large number of tools to accelerate research. It's amazing what technology has suddenly made possible."

Examples of these tools include:

- Mouse models. We have identified existing models in academic labs. We also applied to Jackson Labs Center for Precision Medicine, a leading biomedical institution, for the development of mouse models. They have generously agreed to develop multiple mouse models on a pro bono basis, including humanized mice towards the pursuit of gene therapies.
- Dozens of MED13L cell lines, induced pluripotent stem cells (iPSC's) and neurons.

- Diagnosis code. Leading law firm Hogan Lovells has generously donated their time to help MED13L get an official ICD-10 diagnosis code, with benefits for identifying patient population, educating physicians, and health insurance reimbursement.
- The development of a patient registry and natural history database in order to facilitate clinical trials, as well as an online community forum Cure MED13L.

In addition, the MED13L Foundation has partnered with Rarebase, a public benefit precision medicine company that leverages cutting-edge technology and biology to discover and develop treatments for the millions of people worldwide living with a rare disease. The Rarebase Function™ platform combines leading-edge artificial intelligence and high-throughput assay systems to identify existing FDA-approved drugs that may improve MED13L and that could potentially be repurposed for MED13L patients in the near term. For more information about Rarebase, visit www.rarebase.org.

This summer, the MED13L Foundation is organizing for another in-person gathering of global MED13L families and researchers in Baltimore August 5 – 8, thanks to the generosity of the Simons Foundation, who are organizing and sponsoring the conference for several rare disease communities. CURE MED13L also just announced the formation of its Scientific Advisory Board, including Dr. Wendy Chung of Columbia University, Dr. Chad Grueter of the University of Iowa, Dr. Reza Asadollahi of the University of Zurich, Dr. Randy Strich of Rowan University, Dr. Jennifer Bain of Columbia University, Dr. Jamal Ghoumid of the University of Lille, Dr. Koh-ichi Nagata of Aichi Developmental Disability Center, Dr. Nadav Ahituv of the University of California, San Francisco, and Dr. Christine Gurnett of Washington University in St. Louis, all distinguished leaders in human genetics and the MED13L research field.

Lacey Akins, a MED13L parent and Marketing and Fundraising Coordinator for the MED13L Foundation, notes “we are deeply grateful to these wonderful partners for their support. As our momentum builds, we need to raise significant amounts of capital to fund these research and development initiatives.”

This one-million-dollar fundraising campaign has kicked off with the generous donation of a $100,000 matching grant by a relative of a MED13L family member. This

donor, who has a background of professional success in the gene therapy field, notes: "I have spent time with a large number of rare disease foundations in the span of my career. What CURE MED13L has accomplished in a short period of time and with so little funding has been astounding. We have structured our donation as a matching grant to encourage engagement across the MED13L family community. Commitment from the broader community is key to success. Patient groups that organize together are the ones that are able to change the course of their children's lives — and do it in an accelerated way."

Vanessa Dias, a MED13L parent, Registered Nurse, and Co-Leader of the CURE MED13L Initiative, states that "CURE MED13L is dedicated to making a difference in the lives of those living with MED13L. Advocates for those with rare diseases require strength and passion to push forward novel ideas and research and help shepherd ground breaking therapeutics from the laboratory to clinical trials. Our goal is to push the boundaries of medicine to serve our rare disease community".

About CURE MED13L: Cure MED13L is an initiative of the MED13L Foundation, a non-profit organization founded in 2017, dedicated to supporting families with MED13L and to finding a cure. MED13L is a rare disease that was first identified in 2013 and is a leading cause of autism and intellectual disability and includes other symptoms. For more information, please visit the MED13L Foundation website or the Cure MED13L community forum.

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