ROSAMUND STONE ZANDER TRANSLATIONAL NEUROSCIENCE CENTER

THE MED13L FOUNDATION

MED13L FOUNDATION TO LAUNCH NATURAL HISTORY STUDY OF MED13L SYNDROME WITH BOSTON CHILDREN'S HOSPITAL

Boston Children's Hospital

HARVARD MEDICAL SCHOOL TEACHING HOSPITAL

The MED13L Foundation today announced a nearly $250,000 grant to help launch an innovative natural history study at Boston Children's Hospital's Rosamund Stone Zander Translational Neuroscience Center (RSZ TNC). The MED13L Syndrome Investigation of Natural History and Development (MIND Study) is a three-year research initiative designed to build on existing data and accelerate clinical trial readiness for the MED13L Syndrome community.

This commitment represents a path forward for families affected by MED13L Syndrome, a rare neurodevelopmental disorder, underscoring the MED13L Foundation and Boston Children's shared mission to further understand and drive forward treatments for MED13L Syndrome and other rare neurodevelopmental disorders (NDDs).

“For families affected by MED13L Syndrome, the MIND Study represents both hope and momentum,“ says Kathleen Barry Boychuck, Board Chair of The MED13L Foundation. “Through a strategic collaboration, we are closing critical gaps in what science doesn’t yet understand —gaps that have left our community without answers for far too long. By capturing comprehensive, longitudinal data, we are telling our children’s stories in a way science can recognize, study, and act on. This collaboration marks a foundational step toward understanding MED13L, improving clinical care, and building the path to future treatments.”

rained and a multidisciplinary team of researchers and physicians has been assembled under the RSZ TNC. ACTION has been approved by Boston Children’s Hospital’s Institutional Review Board (IRB) and recruitment for MED13L Syndrome will begin soon.

The MED13L Foundation is the second NDD foundation to join the RSZ TNC's Accelerating Clinical Trial Readiness Innovations for Monogenic Neurodevelopmental Disorders (ACTION) Initiative, an umbrella program designed to establish clinical trial readiness for rare NDDs, leveraging shared RSZ TNC resources and pilot funding. Led by Principal Investigator Dr. Maya Chopra, Clinical Geneticist and Director of Translational Genomic Medicine of the RSZ TNC at Boston Children's Hospital, ACTION provides scalable infrastructure for standardized data collection from individuals with NDDs, customized to the specific needs of individual disorders. A Clinical Trials Readiness Fellow has been hired and trained and a multidisciplinary team of researchers and physicians has been assembled under the RSZ TNC. ACTION has been approved by Boston Children's Hospital's Institutional Review Board (IRB) and recruitment for MED13L Syndrome will begin soon.

“For rare genetic neurodevelopmental disorders, a major barrier to therapeutic readiness is the availability of high quality, rigorous natural history data to inform meaningful clinical trial endpoints,” says Dr. Maya Chopra, ACTION Initiative Principal Investigator and Clinical Geneticist. “The ACTION initiative allows us to address this barrier in a scalable manner. We are thrilled to collaborate with the MED13L Foundation to advance clinical trial readiness for this syndrome.”

In the MIND Study, data from 30 individuals with MED13L Syndrome will be collected annually over three years to better understand disease progression over time and determine meaningful endpoints for future clinical trials. Evaluations will include interviews with the study team, neurological and physical exams, review of medical records, and neurobehavioral assessments. Up to 20 participants will have the opportunity to complete their initial neurobehavioral assessments and visits with the study team during the 2025 Research and Family Meetup being held July 9-12th in Waltham, Massachusetts, where the theme is inclusion and every story matters.

“As a mom to a child with MED13L, this study is more than research—it’s hope in action, “ says Ann Archibald, mom of Molly, and a MED13L trailblazer. “It’s our chance to be seen, to be heard, and to shape a better future for all our children.”

More information on eligibility and enrollment will be shared with the MED13L community in the coming weeks.

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Contact:
Katie Barry Boychuck,
Board Chair
MED13L -The MED13L Foundation Inc.
(773) 629-0521
kbboychuck@med13l.org