ROSAMUND STONE ZANDER TRANSLATIONAL NEUROSCIENCE CENTER

IDEFINE
The Kleefstra Syndrome Foundation

# IDEFINE COMMITS $300,000 TO LAUNCH NATURAL HISTORY STUDY OF KLEEFSTRA SYNDROME WITH BOSTON CHILDREN'S HOSPITAL

Boston Children's Hospital

HARVARD MEDICAL SCHOOL TEACHING HOSPITAL

BOSTON, March 21, 2025 — IDefine – The Kleefstra Syndrome Foundation today announced a $300,000 grant to help launch a groundbreaking natural history study at Boston Children's Hospital's Rosamund Stone Zander Translational Neuroscience Center (RSZ TNC). The three-year study will advance critical research and help prepare the Kleefstra syndrome (KS) community for future clinical trials.

This commitment marks a major milestone for families affected by KS, a rare neurodevelopmental disorder, underscoring IDefine and Boston Children's shared mission to drive forward treatments for KS and other rare neurodevelopmental disorders (NDDs).

"This study is a critical step toward therapeutic development for Kleefstra syndrome," said Geoff Rhyne, CEO of IDefine. "By supporting this work, we are creating a path forward for families who are urgently waiting for treatments and potential cures. The research partnership with Boston Children's Hospital ensures this effort is led by one of the world's foremost research institutions, with deep expertise in rare diseases like KS."

The KS community is the first to leverage the RSZ-TNC's pilot funding of the Accelerating Clinical Trial Readiness Innovations for Monogenic Neurodevelopmental Disorders (ACTION) Initiative, an umbrella program designed to establish clinical trial readiness for rare neurodevelopmental disorders (NDDs). Led by Principal Investigator Dr. Maya Chopra, Clinical Geneticist and Director of Translational Genomic Medicine of the RSZ TNC, 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 KS will begin soon.

In the KS longitudinal study, clinical and biological data from 30 individuals with KS will be collected over three years to better understand disease progression, identify potential biomarkers, and determine meaningful clinical trial endpoints.

“One of the greatest challenges in rare disease research is reaching a state of clinical trial readiness,” said neurologist Dr. Siddharth Srivastava, Director of the IDefine-supported KS Clinic at Boston Children’s Hospital and Principal Investigator of the KS arm of the ACTION study. “This natural history study allows us to gather the vital data needed to design and execute future clinical trials for KS. We are grateful for IDefine’s commitment to advancing this important work.”

Participants with a confirmed molecular diagnosis of KS — either a deletion impacting 9q34.3 including EHMT1, or a pathogenic EHMT1 variant — will undergo annual evaluations for three years. These evaluations include neurological and physical exams, neurobehavioral assessments, EEGs, actigraphy monitoring, and the study of blood samples for potential biomarkers.

The study is also designed to align with care families already receive through the Kleefstra Syndrome Clinic at Boston Children’s Hospital, making it easier for families to participate while receiving ongoing medical support.

“Our community has always stepped up when it matters most,” Rhyne added. “This study will require families to commit to regular visits and assessments, but it’s how we ensure that when a potential treatment becomes available, we are ready to move forward rapidly on a trial. Together, we’re building hope for every child and family affected by Kleefstra syndrome.”

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

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