

# ***Baby and Toddler Participants Needed: Study on KAT6 Disorders***

## **What is this study about?**

This research study aims to examine early developmental profile and trajectory of KAT6 disorders.

## **Why participate?**

- Before clinical trials can be constructed to treat cognitive impairment in genetic diseases, scientists must first understand the cognitive phenotype of the disease in order to identify potential outcome measures, which has to be applicable across age and severity of cognitive impairment.
- Currently, there are limited to no existing literature on early developmental profiles of individuals with KAT6 disorders, including developmental progress or milestones. As such, it is unclear if developmental domains (cognition, language, motor functioning) make gains at different pace, warranting focused domain-specific interventions in early childhood.

## **Who is eligible?**

- Any individual under 5 years of age and is willing to be seen at the Kennedy Krieger Institute outside of the conference dates. We are happy to schedule interested families who are in town for the conference between June 3-6, June 10-14, or at a later date. A copy of the child's genetic test results is required to molecularly confirm the diagnosis.

## **What does it entail?**

- Caregivers will be given an online research intake form and consent form. In addition, a copy of your child's genetic test results is needed to confirm the diagnosis.
- This study requires two in-person visits, one year apart from each other.
  - 1.) In-person testing with your child: Your child will be given a battery of tests to assess early cognition, language and motor skills that will take between 30-90 minutes to complete pending on their age. We will schedule the same testing a year later for your child so we can assess changes in these domains. Part of the in-person visits include a structured interview with parents which will take about another 30 minutes, totaling 60-120 minutes per visit. 2.) Caregivers will be given a battery of questionnaires to complete remotely to index their child's day to day behavioral functioning across both visits.

## **Who do I contact to enroll in this study?**

- If you would like to enroll in this study or have questions, please contact the principal investigator: Rowena Ng ([ngr@kennedykrieger.org](mailto:ngr@kennedykrieger.org)).