

Participants Needed: Study on KAT6 Disorders

What is this study about?

This research study aims to identify the unique cognitive profile 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.
- Given our prior research findings, a new battery focused on nonverbal cognition will be tested to determine its sensitivity in measuring cognitive functioning in KAT6 disorders across different developmental periods and range of severity in intellectual impairment.

Who is eligible?

- Any individual between age 5 to 18 years who is attending the KAT6 Conference in Baltimore (June 8, 2024) or willing to be seen at the Kennedy Krieger Institute. 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 has three components:
 - 1.) In-person testing with your child: Your child will be given a battery of cognitive tests (e.g., paper and pencil or computerized puzzles and games) that will take about 90 minutes to complete. 2.) Your family will be contacted for a follow-up virtual Zoom session to complete a structured interview on your child's communication skills. 3.) Caregivers will be given a battery of questionnaires to complete remotely to index their child's day to day behavioral functioning.

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).
- *Please note, study enrollment and scheduling for this study at the conference are based on a first come first serve basis.*