



A Clinical Study of the Brain and Cognition in Kabuki Syndrome

What: A clinical research study looking at specific neurologic and cognitive characteristics in KS. The purpose of the study is to discover specific areas in which people with KS are different from their peers and use those as targets for treatment. We expect this study to lead directly to a clinical trial to treat the cognitive and behavioral manifestations of Kabuki syndrome.

The study has 2 parts spread over 2 consecutive days:

1. Cognitive and behavioral testing – done one on one with a trained investigator. Involves memory games, puzzles, reading, and questionnaires. Takes about 3-4 hours one day and about 2 hours the other day.
2. MRI – a scan to take a picture of the brain but we are taking a very focused picture of a specific area that we suspect is different in those with KS. The scan takes about 30 minutes but we allot an hour in case breaks are needed. We know that lying still in an MRI scanner can make anyone anxious and claustrophobic so we will have a special 2 hour session either the day before or the morning before the MRI in a mock scanner with trained behavioral specialists to lessen the anxiety and help get through the procedure.

Who: Any person 13 and older with a genetically-confirmed diagnosis of Kabuki syndrome type 1 (mutation in KMT2D)

Where: The cognitive testing and MRI portion will occur at Kennedy Krieger Institute in Baltimore. Participants will come for a 2 day visit.

Why: Mouse models of KS have shown cognitive improvement when treated with certain medications and a specific diet targeting the mechanism of the underlying genetic defect. If we can identify these areas needing improvement in human patients, known in research as outcome measures, then we can move quickly to a clinical trial looking at treatments for the cognitive and neurologic problems of KS based on this work and the work in mouse models.

When: We will begin study visits in January 2021.

How: If interested or if there are further questions, please contact Dr. Jacqueline Harris and/or her research coordinator Jennifer Johnson. At the following phone number or emails:

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The Principal Investigator of this study is Dr. Jacqueline Harris. The IRB number is IRB00240569.

Sample Study Visit Schedule	
Day 1	Day 2
Brief clinical examination (15 min)	Cognitive testing (2-3 hr)
Cognitive testing (2 hr)	
Practice in mock scanner (1 hr)	
Lunch	
MRI in the actual scanner (1 hr)	Cognitive testing (2 hr)