



A Pilot Trial of a Dietary Therapy for Kabuki Syndrome

What: A research study looking at specific neurologic and cognitive characteristics in adults with KS and then determining if there is improvement after 12 weeks of a special diet. The purpose of the study is to determine whether a specific diet may be an effective treatment for KS and lead directly to a possible treatment.

The study involves an initial 2 day visit to Baltimore followed by 12 weeks of a special diet called a Modified Atkins Diet at home with weekly check-ins and then a return visit to Baltimore at the end of 12 weeks.

Initial visit has 3 parts spread over 2 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 & 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.
3. Training session on Modified Atkins Diet lasts 1-2 hours.

12 weeks diet therapy at home:

1. Adhere to the diet and keep daily diet logs
2. Weekly check-ins with investigators
3. Blood and urine testing

Final end of study visit in Baltimore is 1 day:

1. Cognitive and behavioral testing: Repeat of what was done at initial visit

Who: Any adult 18 and older with genetically-confirmed Kabuki syndrome type 1 (mutation in KMT2D)

Where: The initial study visit with cognitive testing and MRI will occur at Kennedy Krieger Institute in Baltimore. Participants will come for a 2 day visit. 12 weeks of diet will occur at home in your normal routine. Return visit to Baltimore will occur at the end of the 12 weeks for a 1 day visit.

Why: Mouse models of KS have shown cognitive improvement when treated with a specific diet targeting the mechanism of the underlying genetic defect. If we can identify areas needing improvement in human patients with KS, known in research as outcome measures, then we can see if the same diet may be effective in people with KS. In short, this may be a potential treatment for the cognitive and behavioral issues in KS!

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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