

The Roya Kabuki Program
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NATURAL HISTORY STUDY OF KABUKI SYNDROME

The Roya Kabuki Program is working towards advancing therapies for patients with Kabuki Syndrome. Consequently we are enrolling children and adults into a natural history study to prepare for clinical trial readiness. The goal of this study is to define neurobehavioral outcome measures (for example, visual spatial skills or anxiety) that may, in future, be used to assess therapeutic efficacy in Kabuki syndrome and help define its natural course.

Participation in this study opportunity generally involves one in-person visit to Boston Children's Hospital for Neurobehavioral Evaluation by Kabuki Clinical Psychologist, Dr. Benjamin Goodlett. This visit will last **approximately 1.5 hours**.

1. The first part is working with the child on standardized activities to understand the child's cognitive development. The tasks might involve drawing pictures, solving puzzles with blocks, and several other activities that are similar to schoolwork like explaining vocabulary words. This portion will take **approximately 30 minutes**.
2. The second part of the evaluation is working with a parent or guardian to complete forms and an interview. This will take **approximately 50 minutes**.
3. There will also be time for discussion of ongoing research efforts and what we are learning about Kabuki Syndrome. Please remember this is a targeted research visit to assess specific areas of cognitive and behavioral functioning for our research goals. This is not a clinical evaluation which is tailored to each child with the goal of providing individual feedback.

There is no requirement to be a patient of Boston Children's Hospital to participate in this study opportunity. Research visits will not be billed to participating families. If you are interested in obtaining a clinical evaluation, this may be facilitated through the Roya Kabuki Program.

Please reach out to program coordinator Tara Daly by email (kabuki@childrens.harvard.edu) or phone (857-218-KBKI) for more details.