

The Roya Kabuki Program
3 Blackfan Circle, CLS 15030 (BCH 3150), Boston, MA 02115
857-218-5254 | kabuki@childrens.harvard.edu

Ophthalmology Study Opportunity

The Roya Kabuki Program is working towards advancing therapies for patients with Kabuki syndrome. As there are many different organ systems affected to various degree, we are particularly interested in studying the natural history of Kabuki syndrome across the entire age spectrum. One focus is on eye disease and eye related findings.

In order to learn more about Kabuki syndrome and the related eye phenotypes, we are collecting records related to eye exams, ophthalmology clinic notes, and other related medical records. Participation in this study opportunity generally involves connecting with the Roya Kabuki Program to provide access to these medical records and completing future related surveys or questionnaires.

You or your child are eligible if the following applies:

- Genetically confirmed diagnosis of Kabuki syndrome
- History of eye findings of any kind
- Documented eye examination and related medical records

If you are interested in learning more or providing records, please reach out to program coordinator Tara Daly by email (kabuki@childrens.harvard.edu) or phone (857-218-KBKI) for more details.

There is no requirement to be a patient of Boston Children's Hospital or travel to Boston Children's Hospital to participate in this study opportunity. Our research would not be possible without the selfless participation on families like yours. We thank you for your time, interest, and participation and look forward to advancing Kabuki syndrome research with you.