

Roya Kabuki Program at Boston Children's Hospital Progress report Mid YEAR 2 (07/18-11/18)



Establish Roya Kabuki Telehealth Clinic for Kabuki syndrome (start 09/18):

Milestone: establish Telehealth clinic

Roya Kabuki Clinic has become increasingly accessible as we launched the Roya Kabuki Telehealth Clinic, a virtual visit option for our patients, on September 1, 2018. By offering virtual visits we can decrease the overall time and costs generally associated with medical appointments to improve health outcomes for our clinic patients. Interactive, real-time appointments can take place wherever patients may have a computer or mobile device with live video capabilities available.

Patient outreach activities (in chronological order):

Milestone: continue patient outreach

July 13-14, 2018 ATK family event in Cleveland, Ohio. Presented information on the established Roya Kabuki Program, actively recruited families to research protocol. 29 new families were consented, 30 families who were previously enrolled in the study were provided with updated study materials. At this event, Dr. Olaf Bodamer was awarded the 2018 Elizabeth Golab Memorial Award from the ATK organization. The award is given to individuals who have a positive impact on the Kabuki Syndrome community and who are passionate about patient care and research.

October 19-21, 2018 Annual Texas Family Gathering. Two members of the team attended the event, presented information on the research and clinical programs, and provided enrollment materials to 15 new families.

October 23, 2018 Kabuki Awareness Day. Halloween card and treats sent to 145 families.

Patient recruitment:

Milestone: Recruit 10-15 families/quarter, build upon Biorepository

The newest version of the study protocol was approved by the BCH Institutional Review Board on July 10, 2018. The program coordinator will facilitate transferring all enrollments prior to this date into this new protocol. All newly recruited families from this time forward will enroll directly into the newest protocol to continue building the biorepository for Kabuki Syndrome data.

	Individuals (index, siblings, parents, etc.)	Families
In Contact	409	157
Consented	266	93

157 families have been in contact with the Roya Kabuki program since September 2017 (start date of the program coordinator Tara Daly), with 70 of these families initiating contact since early July 2018. 93/157 families have been consented to the research protocol. 142/266 blood samples, 119/266 urine samples have been collected and stored at -80°C for analysis. 77/93 index patients have provided skin tissue samples. We have collected muscle tissue biopsies from 2/93 index patients. These samples constitute the present (or current) Kabuki biorepository at Boston Children's Hospital.

Internet Presence:

Milestone: develop non-BCH Webpage

Webpage (www.Royakabuki.org) went live in 11/2018. This page successfully shares the story of the family, the research team and current efforts, and contains a donation portal open to donors worldwide.

Collaborations:

Milestone: solidify Kabuki network

Pre- and Peri-natal History project with Cincinnati Children's Hospital

The Roya Kabuki research team collaborated with Dr. Andrew Lindsley, MD, PhD and clinical fellow Dr. Chen Rosenberg to create the first large dataset on pre- and perinatal history in patients with genetically and/or clinically confirmed cases of Kabuki Syndrome. Research findings were presented at the October 2018 American Society of Human Genetics Annual Meeting (see poster for reference).

Scientific work:

Milestone: not defined

The research team has successfully established fibroblasts with the tissue samples collected thus far. The collected whole muscle tissue samples have been analyzed to identify histological differences between KS and control samples.

Grant submissions:

Midyear 2 Milestone: submit NIH U54 Grant

- 1) NIH U54 Rare Disease Consortium –“Disorders of Histone Modification” (application submitted 10/18). Budget \$5 million (1 million/year for five years). *Decision Pending.*
- 2) Takeda, “Identification of Biomarker”. Budget \$18,000. Approved.

Publications and presentations:

Milestone: not defined

1. Poster presentation at the American Society of Human Genetics Meeting in San Diego. C.E. Rosenberg, T. Daly, C. Hung, E. Gussoni, A.W. Lindsley, O. Bodamer. Pre- and Perinatal History in Kabuki Syndrome