Academic and community collaboration to accelerate Kabuki syndrome awareness, research, and clinical care

Tara Daly MSc *and* Olaf Bodamer, MD, PhD, *with* Rene King, CEO
Division of Genetics and Genomics, Boston Children's Hospital, Roya Kabuki Program *with* All Things Kabuki (ATK)

Abstract

Kabuki syndrome (KS) is a rare congenital disorder. Patients with KS typically require care from a geneticist, along with several additional of specialists through their lifetime. Being a rare disease, most providers including geneticists are not familiar with standard of care and treatment for patients. The Roya Kabuki Program (RKP) at Boston Children's Hospital (BCH) and the All Things Kabuki (ATK) patient advocacy group aim to work together to impact education in the medical and lay communities and pursue research that will improve patient care and health outcomes. Collaboration on the following three key action items: consistent communication, collection of patient feedback to guide research and clinical care goals, and planning of educational awareness events in the medical and patient communities are essential to maximize impact for the KS community. Education, research, and awareness about a rare disease such as Kabuki syndrome is most effectively and appropriately pursued in a joint effort between medical academia and patient advocacy organizations.

Background

Academia:

- The RKP at Boston Children's Hospital has access to the medical and academic communities at Boston Children's Hospital and both national and international institutions.
- The RKP has identified a network of physicians, researchers, and various health care providers from these communities to collaborate and to advance Kabuki syndrome research and clinical care over the past three years.

Community:

- ATK has demonstrated a commitment to advocacy, education, and support amongst the Kabuki syndrome population since their establishment in 2013.
- Social media forums, support programs, and in person family gatherings create bonds that are often otherwise difficult to come across in a rare disease group.

Objectives

The need for greater education and awareness of Kabuki syndrome amongst medical providers, researchers, and the patient communities is not unlike many other rare disorders. Kabuki syndrome is a complex disorder that often requires care from any different specialty departments throughout a child's lifetime. Despite the complexity and intensive care needed there is no targeted treatment or drug therapies specifically for the Kabuki syndrome community.

While the Roya Kabuki Program has established a research and clinical program to aid this, receiving feedback from the patient community to determine must be the ultimate driver to achieving improved health outcomes for the patients.

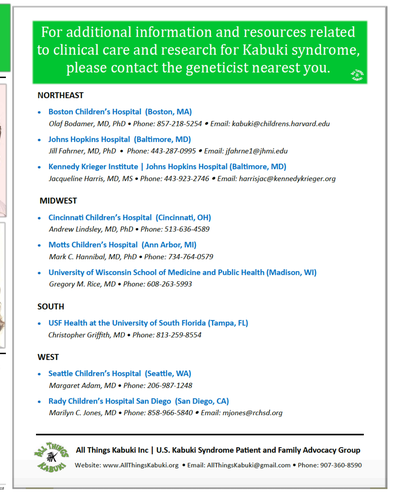
Objectives:

1. Increase education
2. Pursue research

Methods

The RKP has committed to take this journey alongside ATK and the entire Kabuki syndrome patient community. The Roya Kabuki Program and the All Things Kabuki organization actively collaborate on the following three key action items:

1. Consistent Communication: Active lines of communication between the researchers, physicians at BCH and volunteers of ATK will allow for transparency and common goals to be maintained throughout collaboration.
2. Collection of patient feedback to guide research and clinical care goals: ATK's patient community provides critical information to the RKP researchers and providers who are determining care guidelines in the clinic and research strategies and clinical trial end points.
3. Planning of educational awareness events in the medical and patient communities: The RKP and ATK host various events in tandem such as annual family gatherings to promote awareness, creating joint teams for annual charity walks to raise funds and educate others at BCH, and provide research driven updates in a community focused manner.



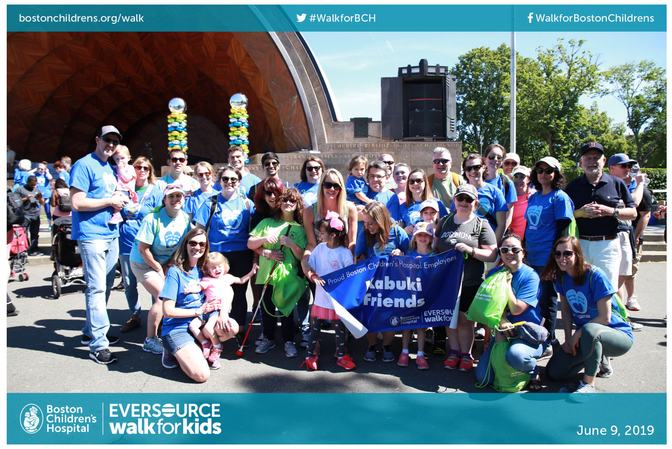
Results

2013: ATK was established by Kabuki syndrome parent Rene King. ATK became an approved non-profit in **2015**. ATK aims to raise international awareness about KS and offer support to families in the patient community.

2017: Following the diagnosis of a newly diagnosed child, Olaf Bodamer established the RKP at BCH with the mission: care, change, cure. The RKP offers both research and clinical opportunities for children and adults with KS.

2017: ATK invited the RKP team to join their annual family gathering. The ***collaboration*** between patient advocacy and academic organizations has been ongoing ever since.

Results



When the RKP and ATK combine the impact on the patient community grows. This has led to changes in clinical structure and direction for research goals.

1. RKP has now dedicated one week each month for comprehensive clinic visits. This change was made after feedback from ATK revealed positive experience families shared when they were able to receive clinical care and meet other families traveling to BCH at the same time.
2. ATK has shared patient questions and topics of interest that have directly led to the creation and distribution of new study surveys to be amongst RKP research participants.
3. The RKP has participated in ATK's Annual Family Gathering since 2017. This provides a unique opportunity for the medical community and the patient community to build a relationship that may be otherwise unattainable in a typical clinic room.
4. Fundraising for rare disease is difficult due to lack of awareness. The RKP and ATK reach a broader audience together when raising necessary funds for community and research efforts during events such as the Eversource Walk for Boston Children's Hospital.

Conclusions

- Collaboration is essential for building an environment that welcomes communication between patients and medical providers. The RKP's continuous participation in community engagement activities such as those led by ATK, amongst others, can build a greater sense of trust that is necessary to invite more participation and sharing of critical patient experiences.
- Given the opportunity to hear directly from a broad patient population, researchers and clinicians learn more about the most immediate and meaningful needs to families. When this information is used to guide initiatives led by the RKP and additional connected academic institutions, the resulting effort will have the greatest possible impact on the KS population.
- Future research will not only uncover new information about KS, it will also have significantly greater impact on quality of life in the overall community than research without consideration of patient input. *This will ultimately serve as a catalyst for forward progress in clinical care and quality of life in KS and related syndromes.*