

# Advancing Kabuki syndrome awareness, research, and clinical care through academic and community collaboration



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## Background

Kabuki syndrome (KS) is a rare and complex genetic disorder requiring specialized care across the lifetime.

All Things Kabuki (ATK) is a KS patient advocacy organization committed to advocacy, education and support for the KS community.

The Roya Kabuki Program (RKP) at Boston Children's Hospital (BCH) is a dedicated program for the advancement of clinical care and research in KS.

2013

2017

## Purpose

There is a greater need for education and awareness of KS among medical providers, researchers, and patient communities.

### Objectives

- Increase provider education on KS and unique patient needs.
- Increase research endeavors and participation.
- Work towards developing targeted treatments and therapies, of which there currently are none.

## Methods

The RKP has identified a network of physicians, researchers, and health care professionals dedicated to advancement of KS research and care.

The RKP and ATK have committed to working together to improve patient care and lives.

- **Consistent communication and collaboration** between the RKP and ATK teams to establish and reach common goals.
- **Correspondence of feedback** has allowed for modifications to clinical and research approaches to better suit the needs of the KS community.
- **Collaboration** on education and research projects such as the patient registry.

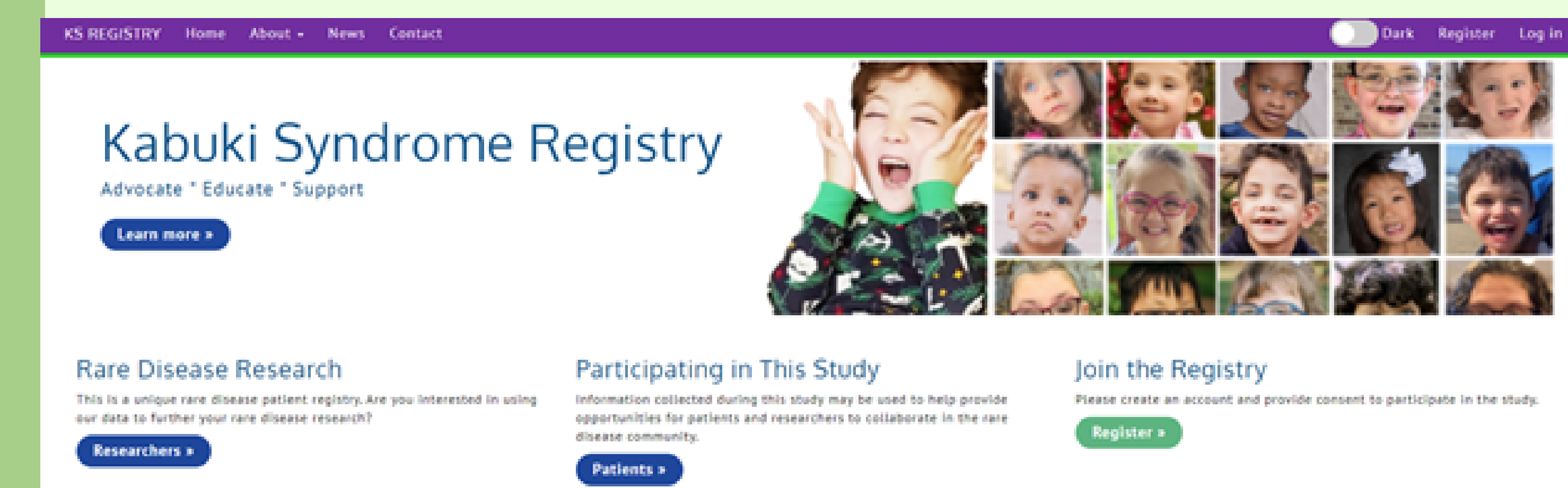
## Results

Collaboration between the RKP and ATK has allowed for greater impact on the KS community. This work has improved patient care through feedback and support from ATK.

- ATK offers social media forums, support groups, and in-person gatherings.
- The RKP hosts "The More You Know" webinar series to keep the KS community informed on research.

## Results

- Dedicated clinic days for KS patients with opportunity for research participation.
- Creation of educational flyers for clinicians.
- ATK-led fund raising efforts to support community and research endeavors through events such as the Eversource Walk for Kids.
- ATK, with support and feedback from the RKP, developed the first Kabuki Syndrome Patient Registry hosted through NORD. This registry will allow patients to easily participate in research endeavors.



## Conclusions

Ongoing collaboration between the RKP and ATK has allowed for improvement in clinical care, research endeavors, and greater awareness of KS. This has laid a foundation of trust within the larger community, which in turn puts patient needs and goals at the forefront of clinical care, research, and advocacy.