ALL THINGS KABUKI

All Things Kabuki is the only U.S. non-profit organization advocating and offering support for Kabuki syndrome.

Originally founded in October 2013, ATK is a volunteer run organization based out of Wasilla, Alaska. ATK’s volunteers are dedicated to raising awareness and offering support to individuals and families affected by Kabuki syndrome.

For more information about Kabuki syndrome, or our organization, please visit our website.

Interested in volunteering? Have a program idea? Want to make a tax deductible donation? Please contact:

Rene King, President & Founder
Email: AllThingsKabuki@gmail.com
Phone: 907.360.8590

Kabuki Syndrome Research @ Johns Hopkins

If you are interested in making a tax deductible donation to the research project run by Dr. Hans T. Bjornsson, Director of Epigenetic and Chromatin Clinic at Hopkins, please visit the following link and designate funds to Kabuki Syndrome Research.

https://secure.jhu.edu/form/igm

Disclaimer: There is no affiliation or endorsement between JHU and ATK.

And now these three remain: faith, hope and love.
But the greatest of these is love. - 1 Corinthians 13:13
**What is Kabuki Syndrome?**

Kabuki syndrome, first described in Japan in 1967, became a formal diagnosis in 1981. Kabuki syndrome was originally referred to as Niikawa-Kuroki syndrome. It would later be referred to as Kabuki Make-Up syndrome (KMS), and then shortened to Kabuki syndrome.

Kabuki syndrome is a rare genetic disorder occurring in approximately 1 in 32,000 births worldwide.

The first gene recognized to be responsible for Kabuki was discovered in 2010 at the University of Washington. There are currently two genes identified as the cause for Kabuki syndrome. Mutations on KMT2D, formerly known as MLL2, are believed to affect as many as 75% of those diagnosed. Mutations on KDM6A are less common and present in approximately 5% of cases. Studies have shown there are likely additional genes yet to be identified. Kabuki syndrome can be clinically or genetically diagnosed by a Geneticist.

Children and adults affected by Kabuki experience a variety of symptoms, some of which may include: mild to moderate intellectual impairment, growth delays, low muscle tone, feeding difficulties, heart defects, cleft palate, skeletal abnormalities, visual and/or hearing impairments, communication delays, and autistic-like behaviors. Kabuki syndrome affects each individual differently.

Kabuki syndrome does not shorten one’s life span, however underlying conditions may.

**Medical Management**

Underlying conditions vary in those diagnosed with Kabuki syndrome. Your child’s primary care provider may consider consulting with the following specialists to identify and address any related conditions.

- Audiology (Hearing loss)
- Cardiology (Heart Defects i.e. Coarctation of the Aorta)
- Dental (Missing, Missshaped Teeth)
- ENT (Cleft Lip/Palate, Eustachian Tube Dysfunction)
- Endocrinology (Hypothyroidism, Growth Hormone)
- Gastroenterology (Feeding Tube, GERD)
- Genetics
- Hematology (Blood Disorders)
- Immunology (Immunoglobulin Deficiencies)
- Nephrology (Kidney Abnormalities)
- Neurology (Developmental Delays, Seizure Disorders)
- Nutritionist (Adolescent Obesity)
- Occupational Therapy
- Ophthalmology (Vision Impairments)
- Orthopedics (Hypotonia, Joint Laxity, Hip Dysplasia)
- Physical Therapy
- Speech Therapy
- Urology (UTI’s, Genital Deformities)

**ATK Programs**

**Pay it Forward Campaign** - Raising awareness for Kabuki Syndrome through Random Acts of Kindness. Order your free cards through our website.

**ATK Birthday Club** - Members receive a personalized birthday card, Kabuki vinyl and a small gift to celebrate their special day!

**ATK Family Directory** - Connecting Kabuki families globally. Enroll today!

**ATK Family Spotlight** - Once a month we shine the spotlight on a Kabuki family and their journey.

**Operation Kabuki Warrior** - Personalized get well cards for our warrior kids. Submit your card request through our website.

**Operation Kabuki Christmas** - Blessing Kabuki families experiencing a financial hardship during the Christmas season.

**Annual Awareness Campaign** - Join our Rare Disease Awareness Campaign every February and our Kabuki Syndrome Awareness Campaign every October.

**ATK Family Mentor Program** - Seasoned families are encouraged to enroll to mentor Kabuki families new to their community. Anyone interested in having a local mentor is also encouraged to enroll.

**ATK Pen Pal Club** - You’ve Got Mail! Connecting Kabuki children & adults through snail mail.