

# ALL THINGS KABUKI

All Things Kabuki is a volunteer run non-profit organization based out of Wasilla, Alaska. Founded in October 2013 and incorporated in 2015, ATK is the only United States patient advocacy group supporting individuals and families affected by Kabuki Syndrome worldwide.

We invite you to partner with us in our advocacy efforts. Together we can change the future for individuals affected by Kabuki Syndrome.

Please visit our website for more information about Kabuki Syndrome, our organization, and active research opportunities.

New to the Kabuki community? Interested in volunteering or partnering with ATK? Contact us today.

Email: [info@allthingskabuki.org](mailto:info@allthingskabuki.org)

Mobile: 907.360.8590 (AKST)

Website: [www.allthingskabuki.org](http://www.allthingskabuki.org)

*Your tax deductible donation makes our work possible!*



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



*All Things Kabuki  
5155 E Capon Circle  
Wasilla, Alaska 99654*



*All Things Kabuki's mission is to raise awareness,  
incite research and support individuals and  
families affected by Kabuki syndrome.*



ATK is proud to collaborate with

Roya  
KABUKI at Boston  
Initiative Children's  
Hospital



AWARENESS, EDUCATION  
AND SUPPORT FOR  
KABUKI SYNDROME



*ATK is a 501(c)3 non-profit patient advocacy organization*

# ✿ ADVOCATE ✿

## 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 ie. Coarctation of the Aorta)
- Dental (Missing, Misshaped 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 (Strabismus, Amblyopia, Coloboma)
- Orthopedics (Hypotonia, Joint Laxity, Hip Dysplasia)
- Physical Therapy
- Speech Therapy
- Urology (UTI's, Genital Deformities)

# ✿ EDUCATE ✿

## 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 Kabuki make-up Syndrome (KMS). It would also be named (and less commonly referred to as) Niikawa-Kuroki Syndrome, after the founding doctors. Kabuki make-up Syndrome would later be 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 impairments, hearing impairments, seizures, 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.

# ✿ SUPPORT ✿

## COMMUNITY PROGRAMS

**BIRTHDAY CLUB** - ATK loves to celebrate Kabuki Kids!

**PEN PAL CLUBS** - You've got mail! The Kabuki and Sibling Pen Pal Clubs relaunched in 2020. Join today!

**FAMILY SPOTLIGHT** - Families share their journey with Kabuki Syndrome. NEW in 2023 ... Sibling Spotlight!

**KABUKI MOM/DAD GROUP** - Kabuki parents and caregivers are invited to join the newly launched parent groups and participate in monthly virtual support sessions.

**OPERATION INSPIRATION** - Women supporting women!

**PATIENT REGISTRY** - Empowering Families to Drive Research! The only global Kabuki Syndrome Patient Registry is now live!

**GREEN STAR FAMILY PROGRAM** - A tribute to honor our fallen Kabuki Warriors.

**PAY IT FORWARD CAMPAIGN** - Raising awareness for Kabuki Syndrome through random acts of kindness!

**ANNUAL AWARENESS CAMPAIGNS** - Join our Rare Disease Awareness Campaign every February and our Kabuki Syndrome Awareness Campaign every October on Facebook and Instagram.

**KABUKI COFFEE HOUR** - A one-hour, topic specific, virtual event to engage, support and share.

**OPERATION KABUKI CHRISTMAS** - Blessing Kabuki families experiencing a financial hardship during the Christmas season. Kabuki angel, Elizabeth Golab (1998-2015), was the inspiration behind what began as a random act of kindness and evolved into a patient advocacy organization.

Visit the **PROGRAMS** section of our website for more information about active programs and virtual events.