

Kabuki Syndrome Services

at Kennedy Krieger Institute

Who We Are

Kabuki syndrome is a rare congenital disorder characterized by distinctive facial features, growth delays, mild-to-moderate intellectual disability, skeletal abnormalities and short stature. The syndrome's true incidence rate is unknown, as many people are not diagnosed properly, but is estimated to be 1 in 32,000 individuals.

Kennedy Krieger Institute, through its Neurology Clinic and along with partnering institution The Johns Hopkins Hospital, is one of the few health care providers in the world that diagnoses and treats this syndrome. We provide comprehensive medical evaluations, clinical care and genetic testing. In addition to our expertise in this very rare disorder, our vast resources mean that our patients' needs can be met at one location. It is our goal to help our patients and their families gain a better understanding of their condition and access appropriate medical management to improve their overall health and quality of life.

Who We Serve

Kennedy Krieger provides expert diagnosis and treatment for children, adolescents and adults with Kabuki syndrome. We provide one-time evaluations for diagnostic and management purposes as well as long-term treatment and care.

The disorder affects males and females, and it can affect different organs and cause a variety of problems that our team is able to treat and address, including:

- Growth deficiency
- Feeding difficulties
- Low muscle tone in early years
- Seizures
- Hearing issues
- Cardiovascular abnormalities
- Orthopedic problems

Our Team

Kennedy Krieger is home to a unique collaboration of experts in Kabuki syndrome from various disciplines. Our clinicians, research scientists and rehabilitation professionals work together to provide optimal interdisciplinary care for patients. Because Kabuki syndrome can affect many different systems in the



body, treating a patient with the disorder often requires the collaboration of a wide range of medical specialists. Key specialists who see our patients include those in the following fields:

- Audiology
- Behavioral psychology
- Cardiology
- Genetic counseling
- Neurodevelopment pediatrics
- Neurology
- Occupational therapy
- Physical therapy
- Speech and language therapy
- Orthopedics

We are all born with great potential.
Shouldn't we all have the chance to achieve it?

Our Approach

Evaluation and Diagnosis

Clinical genetic testing for Kabuki syndrome is available and done in partnership with the Epigenetics and Chromatin Clinic at The Johns Hopkins Hospital. If a genetic diagnosis has not yet been made, a genetic counselor guides patients and their families through the diagnostic process.

Treatment

There is no specific treatment for Kabuki syndrome. Instead, treatment is applied to ameliorate specific symptoms in each individual. Treatment may require the coordinated efforts of a team of specialists. Early intervention with education and support services, as well as physical and occupational therapy for improving tone, are critical to ensure that children with Kabuki syndrome reach their potential.

Additionally, we can also facilitate access to Kennedy Krieger's comprehensive team of specialists to assist with specific educational, behavioral and other therapy needs.

Our Research Initiatives

Our laboratory and clinical research programs play an important role for Kabuki syndrome patients. We are dedicated to finding and developing new treatments for this disorder. We have several active studies looking at the neurologic and developmental aspects of Kabuki syndrome.

We maintain strong collaborations with parent and patient advocacy groups throughout the U.S. and Canada. We will continue to dedicate our research efforts toward these goals.

Contact Information

Kabuki Syndrome Services at Kennedy Krieger Institute

For more information, call **443-923-9400**, or call toll-free at **888-554-2080**. TTY users, please contact us at **443-923-2645**, or dial **711** to make a Maryland Relay call.

KennedyKrieger.org

Physicians and Healthcare Professionals

To make a referral, call our Physician Referral Line at **443-923-9403**.

Clinic Location

801 North Broadway
Baltimore, MD 21205

For information about ongoing research studies, please contact **443-923-2746**.

Kennedy Krieger Institute recognizes and respects the rights of patients and their families and treats them with courtesy and dignity. Kennedy Krieger Institute provides care that preserves cultural, psychosocial, spiritual and personal values, beliefs and preferences. Care is free from discrimination based on age, race, ethnicity, religion, culture, language, physical or mental disability, socioeconomic status, sex, sexual orientation, and gender identity or expression, including transgender. We encourage patients and families to become active partners in their care by asking questions, seeking resources and advocating for the services and support they need. © 2017 Kennedy Krieger Institute 06/2017



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