

## DEVELOPMENT AND THERAPIES

Individuals with KS often exhibit delays in multiple areas of development. It is important to have prompt evaluation as early intervention is key. During school years a majority will require appropriate therapies to optimize learning.

Ongoing assessments and interventions are likely for most; enabling positive, productive experiences throughout life.

*Early Intervention* - Identify needs, develop intervention plan, provide therapeutic services, and track progression, optimizing development.



*Physical* - Develop gross (large) motor abilities, improve strength, and assess need of modifications and adaptive equipment for school/home/work.

*Occupational* - Develop fine (small) motor skills, strength, and dexterity. Assess need of assistive devices and adaptations for school/home/work.

*Speech* - Improve articulation, develop language and, if indicated, introduce sign language. Develop oral motor control, especially if tube fed.

*Sensory Integration* - Improve sensorimotor, self-regulation, environmental settings (classroom/home/etc), social skills, and self-esteem issues of sensory integration dysfunction.

*Music* - Many individuals with KS enjoy music and rhythm - an interest that can enhance and motivate learning in many other areas.

## LIVING WITH KABUKI SYNDROME

Kabuki Syndrome is a life-long disability. Although KS is not a progressive condition, new disorders may arise over time.

The level of independence for adults with Kabuki will vary depending upon their individual abilities and degree of family and community support. Living and employment settings are likely to include a variety of options ranging from supervised to independent. It appears the majority will require supervised settings, many will manage with a mixed degree of independence and assistance, and a few may be able to function independently at work and home.

## KABUKI SYNDROME NETWORK

- Kabuki Syndrome Network (KSN) website offers in-depth clinical information, up-to-date syndrome developments, and quality related resources. Website pages are currently available in English, and the gradual development of Spanish pages.
- Connect with others through KSN's email-list discussion group. This very active group has a high rate of participation, and is an invaluable source of emotional and practical support. Simple registration is required.
- Attend/organize get-togethers with other Kabuki families in your area.
- Become a user of KSN's website to access its resources:
  - *Family Directory*: Contains location and contact information of families
  - *Kabuki Journal*: Read archived newsletters



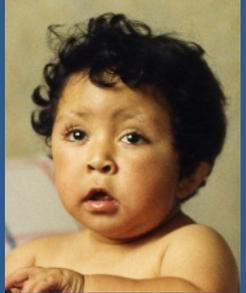
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*The KSN logo (brochure front) is the Japanese character 'K' representing the word 'Kabuki'. Its beautiful form is especially appropriate as it also illustrates two outstretched arms, symbolic of the invitation to network with one another. (Designed by Jos Vergouwen, of the Dutch Netwerk Kabuki Syndroom)*



	<p><i>Diagnostic Criteria &amp; Related Conditions</i></p> <p><i>Medical Evaluation Guidelines</i></p> <p><i>Network Resources</i> <i>* Contacts * Support</i></p>
<p><b>D</b>are to Dream <b>C</b>ourage to Adapt <b>W</b>isdom to Rejoice</p>	
	<p><b>Kabuki Syndrome Network</b></p> <p><i>A global network that promotes and facilitates:</i></p> <p><i>*expanding knowledge of Kabuki syndrome</i> <i>*family support through shared experiences</i></p>



## WHAT IS KABUKI SYNDROME?

First described in 1981 by two Japanese doctors (Niikawa and Kuroki) and named for the likeness of facial characteristics, especially eyes, to the makeup of Japanese Kabuki actors. Kabuki Syndrome (KS) is a congenital multiple-malformation syndrome.

**DIAGNOSTIC CRITERIA:** Diagnosis is based on the recognition of four (*out of five*) main characteristics, with the distinct facial features being imperative. Analysis of chromosomes will typically yield normal results.

- Facial features: long palpebral fissures with eversion of outer third, arched eyebrows with sparse outer half, prominent eyelashes, prominent and/or misshapen ears, and depressed nasal tip
- Skeletal abnormalities
- Dermatoglyphic abnormalities
- Intellectual disability (mild to moderate)
- Postnatal short stature

**ADDITIONAL FEATURES:** Kabuki is a complex syndrome with many associated findings. Co-existing conditions support a diagnosis but are not, separately, considered cardinal traits. Partial list:

- Hypotonia
- Feeding difficulties
- Recurrent infections
- Congenital heart defects
- Renal (kidney/urinary tract anomalies)
- Small mouth, micrognathia (smallness of the jaws), cleft or high arched palate
- Hypodontia (missing teeth), unusually shaped teeth, and misalignment
- Birth - normal weight; infancy & childhood - underweight; pre-teen onward - *possible* obesity
- Early breast development (girls)
- Hearing impaired and/or inner ear malformations

**Diagnostic Note:** The occurrence of associated conditions varies in number and degree. Each individual presents a unique clinical picture.

## KABUKI 'BASICS'

The discoveries of KMT2D gene mutation in approximately 70% of individuals, and KDM6A gene mutation in 9% of individuals, have made it possible to definitively diagnose a significant portion of children. Since it is suspected that numerous gene mutations are responsible for KS, studies are underway in the hopes of further clarifying the genetic causes. It is important to consult your geneticist for up-to-date information on the genetic implications for your family.

As a syndrome, there is no cure. However, many of the conditions associated with KS can be corrected or alleviated with appropriate intervention.

Worldwide, the growing awareness of Kabuki Syndrome is promoting interest within the medical community and generating increased research.

## DIAGNOSIS: ADVISED EVALUATIONS

**Genetics** - Your geneticist can provide ongoing information regarding Kabuki's genetic implications for your family, recent clinical findings, and pertinent genetic studies.

**Cardiology** - If a heart defect is not detected at birth, a referral should be made for assessment of possible cardiac anomalies.

**Ear, Nose and Throat (ENT)** - Frequent ear infections, cleft or high arched palate, hearing loss, and inner ear malformations require evaluation and continued care by an ENT.

**Audiology** - There is an increased risk of hearing loss (conductive and sensorineural). Hearing should be thoroughly evaluated at birth and periodically thereafter.

**Ophthalmology** - Thorough, routine eye exams are recommended due to frequent strabismus, nystagmus, ptosis, coloboma and other defects.

**Orthopedics** - Joint laxity, foot/ankle/gait issues, hip/*patella* dislocation, rib/vertebral and other skeletal anomalies are frequent finding that require orthopedic evaluation and care.

**Dentistry** - Missing, unusually shaped, widely spaced (or crowded), and misaligned teeth are common dental issues. Sensitivity to oral stimulus frequently prevents proper oral hygiene. Early and routine treatment from a pediatric dentist is recommended.

**Urology** - Due to an increased risk of urinary tract malformations, an abdominal ultrasound is recommended.

## RELATED CONDITIONS: EVALUATIONS

**Immunology** - Abnormal immunity test results (particularly hypogammaglobulinemia and IgA), recurrent infections, and allergies should be assessed by an immunologist.



**Hematology** - Autoimmune conditions such as idiopathic thrombocytopenic purpura and hemolytic anemia are associated with KS>

**Endocrinology** - Hypoglycemia, growth hormone deficiency, hypothyroidism, early breast development (girls), and adolescent obesity are possible conditions.

**Feeding and Gastroenterology** - Infant/childhood feeding difficulties (some requiring tube feeding) are common and may persist. Chronic constipation and/or diarrhea are prevalent. Sensitivity to oral stimulus frequently interferes with eating. Assistance to maintain adequate nutritional intake is often necessary.

**Neurology** - Individuals with Kabuki have an increased risk of abnormal brainwave activity (EEG) and seizures. Dimple or sinus in tailbone/sacral areas is common. Structural brain abnormalities have been reported.

**Anesthesiology** - Certain physical (structural) features associated with Kabuki could complicate the effects of anesthesia. Glucose monitoring during all surgeries is important for tube-fed children in case of undiagnosed hypoglycemia.

**Behavior** - Anxiety, attention problems, obsessive-compulsive traits, and autistic behaviors are often observed. There is an increased need for structure. Consultation regarding appropriate educational services, counselling, and/or medication management is important.

*This brochure provides only abbreviated material regarding diagnostic and management guidelines for Kabuki Syndrome. Consult medical professionals and additional sources for more extensive syndrome information.*

*Comprehensive data available on Kabuki Syndrome Network website: [www.kabukisyndrome.com](http://www.kabukisyndrome.com)*