

Kabuki syndrome (KS) *at a Glance*

Kabuki syndrome (KS) consists of differences in facial features and skeleton, persistence of fetal fingertip pads, mild to moderate intellectual disability, and growth deficiency. The condition was first named for the appearance of the facial features, especially the eyes, thought to resemble the make-up used in Japanese Kabuki theatre.



Meet Susanne on page 9.

Things to Think About

1. Medical / Dietary Needs

What you need to know

- No special diet is required for KS although a well-balanced diet is important. Certain children may have a GI tube or medications/accommodations for reflux.
- The severity of any one of the possible medical problems varies widely between individuals. Therefore, it is important to ask the parents about the medical issues in their child.
- School age children with KS may have annual doctor and specialist visits to monitor medical conditions.
- Be aware, or ask a parent, if the child has a medical alert bracelet.

Physical characteristics and/or symptoms:

Not all people with Kabuki syndrome have all of these characteristics.

- Typical facial features in KS
 - Larger appearing eyes due to a longer lid opening
 - Outer part of lower eyelid turned inside out

- Arched and broad eyebrows
- Large, prominent or cupped ears
- Minor skeletal differences
 - Spinal column
 - Butterfly vertebrae
 - Scoliosis
 - Short fingers and toes
 - Curved 5th finger
- Persistence of fetal fingertip pads
 - Usually, these finger pads normally occur in human fetuses but disappear before birth.
- Mild to moderate intellectual disability
- Growth deficiency (35-81%)
 - Small head size which may/may not accompany short stature.

Additional findings

- Congenital heart defects (40-50%)
- Genitourinary differences (25%)
 - Abnormal kidneys or urinary tract
- Cleft lip and/or palate (33%)
- Gastrointestinal problems
 - Anal atresia (absent or narrow opening in the anus)
 - Reflux
 - Diarrhea
 - Constipation
 - Feeding difficulties
- Eye findings
 - Eye lid droops
 - Crossed eyes
 - Coloboma (an abnormal hole present from birth in one of the structure of the eye)
 - Abnormal blue color of the eye lining
 - Abnormally small eye(s)
 - Dry eyes related to sleeping with eyes partially open
- Dental
 - Widely spaced teeth
 - Missing teeth

- Irregular shape and/or misaligned teeth
- Oral sensitivity which may make dental care difficult
- Skin and nails
 - Hyperelastic skin
 - Hands feel soft
 - Nails: absent, incomplete, or fragile
 - Hair differences (i.e. texture)
- Infections and autoimmune disorders more likely
- Seizures (10-39%)
 - Usually controlled with medicine
- Endocrinology abnormalities
 - Premature breast development
- Small mouth, small jaw
- Hearing loss 40%-82% (conductive) and ear pits
- Low muscle tone (25-89%) and loose joints (50-75%)
- Autism or autistic-like features

What you can do

- A yearly checkup and studies as needed should occur in the child's Medical Home.
- Eye and hearing exams may be done at school
- Be aware of any changes in behavior or mood that seem out of line with the situation and notify the parents.
- It is important to be aware of any academic changes. Contact parents when any differences are noticed.

2. Education Supports

What you need to know

It is important to have **HIGH LEARNING EXPECTATIONS** for children who have Kabuki syndrome. Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

Individuals with Kabuki syndrome may have:

- Mild to moderate intellectual disability
- Delays in speech and gross and fine motor skills

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- Weakness in visual spatial skills
- Relative strengths in verbal and non-verbal reasoning

If child has hearing loss, they may need a classroom sound-field FM system.

Many of the speech delays are due to low muscle tone, poor coordination, and facial structure differences. Receptive language is often better than expressive language.

- Articulation errors common because of low oral motor tone and poor coordination
- Abnormal quality of the voice
- Abnormal prosody (rhythm, stress and intonation of speech)
- Possible problems with grammar
- May use gestures instead of words

What you can do

Speech Therapy

- Improve articulation
 - Develop language
 - Teach learning strategies for non-verbal expression.
 - Consider new technology, computers, and possibly sign language depending on fine motor skills. Focus on non-verbal methods of communication.
 - Use augmentative communication aids such as picture cards or communication boards early.
 - Communication should work with child's desire to socially interact with others in functional and natural settings.
 - Make sure children have opportunities for choice and control in their lives (e.g., choose books, colors, food, clothing, play, play partner, etc.).
 - Incorporate typically developing peers into their therapy to promote social interaction as well as provide typical models of language.
- Creating an “AAC Team” can be crucial to a child’s success with a device in an environment such as the school system.
- Find AAC system that allows for maximal social reciprocal communication.
- Encourage finger pointing in early child hood to help with device use as they age.
- Anyone interacting with the child should have education and training on how to encourage reciprocal communication with the device.
- Modeling is important for the use of the AAC device and to encourage its use.
- Continue with strategies that improve oral control to maximize their potential as oral speakers.
- Developing oral motor control is crucial if the child is being tube fed. Oral strengthening exercises as well as sensory and tactile stimulation of the oral musculature can be beneficial for

- acquiring limited verbal output.
- Sensory integration
 - Improve sensorimotor skills
 - Work on self-regulation, social skills, and self esteem issues associated with sensory integration dysfunction.
- Movement
 - Physical, speech, and occupational therapy to enable walking, proper positioning, hand use, communication needs, etc.
 - Ensure are areas are safe, free of obstacles.

Physical Therapy

- Develop gross motor abilities
- Improve strength
- Assess modifications and adaptive equipment for school and home.
- Sensory integration
 - Improve sensorimotor skills
 - Work on self-regulation, social skills, and self esteem issues associated with sensory integration dysfunction.
- Movement
 - Physical, speech, and occupational therapy to enable walking, proper positioning, hand use, communication needs, etc.
 - Ensure are areas are safe, free of obstacles.

Occupational Therapy

- Fine motor
- Strength
- Dexterity
- Assistive devices and adaptations
- Sensory integration
 - Improve sensorimotor skills
 - Work on self-regulation, social skills, and self-esteem issues associated with sensory integration dysfunction.
- Movement
 - Physical, speech, and occupational therapy to enable walking, proper positioning, hand use, communication needs, etc.
 - Ensure are areas are safe, free of obstacles.

3. Behavioral and Sensory Support

What you need to know

People with Kabuki syndrome have a higher incidence of anxiety, attention problems, obsessive-compulsive traits, and autistic behaviors. However, although they may have autistic like behaviors and/or sensory processing disorder, many will not be officially diagnosed with autism.

Possible Communication Issues

- Language delays
- Difficulty waiting their turn
- Interruptions
- Self-talk
- Repetition of questions

Possible Social Interaction Issues

- Poor eye contact
- Understanding social cues
- Abstract thinking
- Short attention span

Sensory Challenges

- Extra sensitive to touch and/or visual stimuli
- May dislike loud noises, smells, food tastes, and textures

Behaviors

- Self-stimulation behaviors
 - Hand flapping
 - Head shaking
 - Rocking
 - Repeating phrases
- Self-injurious behaviors
 - Head banging
 - Biting self

- Very oral (biting on non food items)
- Music
 - Many enjoy music and rhythm
- Play
 - Like to play or do the same thing over and over
 - Enjoy playing with younger kids

What you can do

- Many children with KS will sometimes exhibit some autistic-like behaviors
- Whether or not they have an autism diagnosis, the types of interventions and therapies may be the same
- Structure, routine, and preparation for change are all helpful
- Calming activities
- Clear concrete plans and visual cues (i.e. visual sign for quiet)
- Proactive behavioral plans that include goals, rewards, and consequences for appropriate behavior
- Seating in back section of room and/or allow seating near exit.
- Structure and predictability
- Reduced level of environmental noise/sound, natural lightening, and avoidance of crowded areas.
- Predictable transitions and signal with visual cues
- Non-verbal cues and feedback
- Role playing the behavioral consequences
- Alternative to stressful events
- Breaks and downtime if needed
- Work on conversational skills and friendships

4. Physical Activity, Trips, Events

What you need to know

- Obesity can be problem during puberty
- Any change in routine may produce anxiety, fears, and/or worry.
- If child has any sensory, hearing, or vision issues, may need preferred seating.

- If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming (www.nepassage.org).

What you can do

- Exercise is important and should be encouraged, especially those exercises that build muscle mass and motor skills.
- Coordination may be a problem and make individuals a target for teasing.
- Practice can help.
- Lack of stamina and energy levels can be problem
 - Individuals may not know how to pace themselves or when to rest.
- Keep active!
- Offer anticipatory guidance and preparation to prepare for a change in routine such as a field trip.
- Create a picture story about the upcoming event. The child can rehearse it alone or with others.
- Offer supports as needed for vision and hearing issues.

5. School Absences & Fatigue

What you need to know

- Children with Kabuki Syndrome may be absent due to illness and/or medical appointments. Help to make transitions in and out of school as seamless as possible.
- Individuals with Kabuki Syndrome should not have excess fatigue.

6. Emergency Planning

What you can do

- Develop an emergency plan if necessary, depending on the needs of individual children.

7. Resources

Kabuki Syndrome Network

The Kabuki Syndrome Network strives to support families and professionals to aid in the healthy development of our children. Their pages include especially helpful information on types of therapy used in conjunction with Kabuki and transition. www.kabukisyndrome.com

GeneReviews: Kabuki Syndrome

Learn more about the genetics of Kabuki Syndrome here!

<http://www.ncbi.nlm.nih.gov/books/NBK62111/>

Classroom Accommodations for Students with Visual Issues

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

Genetics Home Reference

Consumer-friendly information about human genetics from the U.S. National Library of Medicine <https://ghr.nlm.nih.gov/condition/kabuki-syndrome>

8. Meet a Child with Kabuki Syndrome

Susanne, the Social Butterfly



Susanne is an 11-year-old girl who lives in Texas with her mom, Susan and family. “I call her my little secretary because she has a memory that won’t quit!” remarks Susan. “She remembers everyone’s birthdays and phone numbers, and you better remember if you tell her you’ll do something on a certain day because she’ll hold you to it!” Susanne is a very sociable, affectionate and outgoing girl. She likes to interview everyone she meets, and will offer up some information about herself as well – like that she’s the youngest of six children!

In the morning, Susanne is very independent and will get herself dressed and ready for school, although Susan admits that she may need to spruce up her hair a bit. She takes the bus and arrives at elementary school, ready for a day of sixth grade. In the mornings, she attends an adaptive dance class that she loves. Her day then consists of regular education classes of science and math, and individualized reading instruction. She also sings in

the school choir. Once per week Susanne works on pronunciation speech sounds in speech therapy. In a recent parent-teacher conference, the team celebrated that Susanne has been doing very well in school and they agreed to create more challenging work with increased responsibilities in school.

Outside of school, Susanne is very popular. The family takes bets that they will not be able to get through a visit at the grocery store or dinner at a restaurant without someone saying, “Hi, Susanne!” She is also very involved at her local church, which she attends every Sunday with her grandparents. Susan notes how much the older people adore her. She is part of a youth group at the church and goes on trips to sing at nursing homes with the youth group.

Susanne’s three oldest siblings have moved out of the house, but Susan says they dote over her, and keep in touch with her often. She has an older brother and sister who still live with her, and in contrast, have more of a typical sibling relationship with Susanne! Susan says they will argue and tell her she’s annoying – typical sibling stuff – but are sympathetic and supportive when Susan has medical flare ups.

Susanne started life in the NICU, born just a little premature. In utero, an ultrasound showed that one kidney was bigger than the other. Other than that, the family did not know until she was born that anything was different about her. When she was born, however, she was only four pounds and it was found that she had only one kidney, a heart defect, and a tethered spinal cord. The heart defect has since resolved, and Susanne had back surgery to fix the spinal issues. Around five years of age, the neurologist suspected Kabuki Syndrome, and genetic testing was done to confirm the diagnosis. At her last appointments, her heart and kidneys have checked out as healthy. She did not have cleft palate, but she did have a high palate. The family has worked with an orthodontist with palate expanders and braces to fix this problem. She has had eye surgery for lazy eyes and ear surgery for cholesteatoma, which left her with decreased hearing in one ear. Susanne does suffer from back pain when walking and gets hip injections. A surgery is being considered for when she is done growing.

Susanne has an active life at school, with her large family, and at church. She goes to birthdays with her friends, and they have come to her birthdays for a sleepover. She even has a boyfriend at school! She has attended two middle school dances this year, and did not want to leave the Valentine’s Day dance! When she grows up, Susanne says she would like to be a teacher.

Advice to Teachers:

- Always have patience.
- Find what the child is capable of and follow their lead.

Advice to Parents:

- Advocate for your child to be in programs that will foster growth.
- Nobody knows exactly what to expect with Kabuki, getting connected with other families can be helpful.