Genetic Education Materials for School Success

Kabuki Syndrome

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Acknowledgment:

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Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

Kabuki Syndrome

At a Glance

Kabuki Syndrome (KS) is a rare genetic condition. It was named for the appearance of the facial features, especially the eyes, thought to resemble the make-up used in Japanese Kabuki theatre. Kabuki Syndrome can affect many parts of the body often causing growth problems, distinctive facial features, and mild to moderate intellectual disability. Most case of KS are the result of new genetic mutations not seen in other family memebrs, but occasionally an affected person is believed to have inherited the variant from one affected parent. Presentation varies significantly from one child to the next.

Meet Susanne on page 11

Common Features of Kabuki Syndrome:

Unique and distinct facial features

- Broad, large foreheads with sparse, arched eyebrows
- Heavy, thick eyelashes
- Wide- set eyes
- Flattened nose
- Large ears

Growth Problems

- Delayed growth – small head, small stature
- Short fingers, especially the pinky
- Misshaped bones in spine – scoliosis or spina bifida possible

Medical complications

- Recurrent ear infections
- Heart defects
- Kidney problems, urinary tract infections
- Possible feeding issues
- Seizures

Cognitive issues

- Developmental delay
- Language difficulties
- Delayed speech
- Autism spectrum disorder

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In Depth

Medical and Dietary Considerations

What you need to know

The severity of any possible medical problems varies widely between individuals. Therefore, it is important to ask the parents about the medical issues in their child.

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
- Growth deficiency - small head size which may/may not accompany short stature.
- Minor skeletal differences
    - Abnormally shaped vertebrae
    - Scoliosis
    - Short fingers and toes
    - Curved 5th finger
- Persistence of fetal fingertip pads
    - Fleshy finger pads that normally disappear before birth.
- Mild to moderate intellectual disability

Medical findings that may occur

- Congenital heart defects
- Genitourinary anomalies
    - Abnormal kidneys or urinary tract
- Cleft lip and/or palate
- Gastrointestinal problems
    - Anal atresia (absent or narrow opening in the anus)
    - Reflux
    - Feeding difficulties
    - Diarrhea and/or constipation
- Eye findings
    - Eye lid droops
    - Crossed eyes
    - Coloboma – missing portion of the eye
    - Abnormal blue color of the eye lining
    - Abnormally small eye(s)

  - Dry eyes related to sleeping with eyes partially open
- Dental
  - Small mouth, small jaw
  - Widely spaced teeth
  - Missing teeth
  - Irregular shape and/or misaligned teeth
  - Oral sensitivity which may make dental care difficult
- Infections and autoimmune disorders more likely
- Seizures Usually controlled with medicine
- Endocrinology abnormalities
  - Premature puberty for females
  - Increased risk of hypothyroidism or low blood sugar
- Hearing loss
- Low muscle tone and loose joints
- Autism or autistic-like features

What you can do

- A yearly checkup and studies as needed should occur in the child's Medical Home. Request copies of any updated plans and review at least yearly.
  - Inquire about any cardiac concerns, seizure medications, feeding issues or other medical concerns specific to the child.
- Encourage frequent handwashing in the classroom.
- Schedule eye and hearing exams at school to minimize absences
- For feeding issues, consider
  - Thickened feedings and positioning after meals to treat gastroesophageal reflux may be beneficial
  - A gastrostomy tube may need to be placed if feeding difficulties are severe.
- Be aware of any changes in behavior or mood that seem out of line with the situation and notify the parents.
- Be aware of any academic changes. Contact parents when any differences are noticed.

Education Supports

What you need to know

It is important to have high learning expectations for children who have Kabuki Syndrome utilizing their strengths and interests as a foundation. Encourage use of the core educational curriculum and modify it to meet the child's individual needs.

Individuals with Kabuki syndrome may have:

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

- Weakness in visual spatial skills
- Relative strengths in verbal and non-verbal reasoning
- Speech delays
    - Many of the speech delays are due to low muscle tone, poor coordination, and facial structure differences.
    - Articulation difficulties
    - Abnormal quality of the voice
    - Abnormal prosody (rhythm, stress and intonation of speech)
    - Possible problems with grammar
    - Receptive language is often better than expressive language.
    - May use gestures instead of words

What you can do

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

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 (AAC) such as picture cards or communication boards early.
    - Communication should work with the 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 and provide typical language models.
- 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 childhood 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.

Physical Therapy/OT

- Follow their recommendations to
    - Develop gross motor abilities
    - Develop fine motor skills
    - Improve strength and dexterity
- Enable walking, proper positioning, hand use, communication needs, etc.
- 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.
- Ensure areas are safe, free of obstacles.

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. Any change in routine may produce anxiety, fears, and/or worry. However, although they may have autistic like behaviors and/or sensory processing disorder, many will not be officially diagnosed with autism. Whether or not they have an autism diagnosis, the types of interventions and therapies may be the same.

Possible Social Interaction Issues

- Poor eye contact
- Problems understanding social cues
- Problems with 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

- Structure and routine
- Predictable transitions and signal with visual cues
- Prepare student for any anticipated changes
- Calming activities
- Alternative to stressful events
- Breaks and downtime if needed
- Clear concrete plans with visual cues (i.e. visual sign for quiet)
- Proactive behavioral plans that include goals, rewards, and consequences for appropriate behavior
- Role playing the behavioral consequences
- Non-verbal cues and feedback
- Work on conversational skills and friendships
- If the child has any sensory, hearing, or vision issues, they may need preferred seating.
    - Seating in back section of room and/or allow seating near exit.
- Reduced level of environmental noise/sound, natural lighting, and avoidance of crowded areas.

Physical Activity, Trips, Events

What you need to know

Support child’s participation in activities with their classmates. Exercise is important and should be encouraged, especially those exercises that build muscle mass and motor skills. Any change in routine may produce anxiety, fears, and/or worry.

What you can do

- Encourage a supportive environment among the students. 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. Remind them to take rest breaks.
- 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.

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 be reminded to pace themselves during physical activity. Fatigue should do pose significant issues during the school day.

Emergency Planning

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

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Resources

Medline Plus

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide a good medical overview of the condition without being too technical.
https://medlineplus.gov/genetics/condition/kabuki-syndrome/

Kabuki Syndrome Foundation

The Foundation's mission is to advocate for individuals with Kabuki syndrome by finding treatments that will improve their lives. KSF drives patient-centered research through fundraising and collaboration with partners across the globe.
https://www.kabukisyndromefoundation.org/

Info for School Nurse and Primary Care Staff

Kabuki Syndrome is a complex condition that will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student’s team.

Gene Reviews is a peer-reviewed point-of-care reference accessible via the National Library of Medicine. The entry for Kabuki Syndrome provides a comprehensive summary of the condition and describes some ongoing surveillance and management issues.
http://www.ncbi.nlm.nih.gov/books/NBK62111/

HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, ensure that the parents have signed a release of information form at the primary care office. The school nurse should keep a copy of that release in the student’s file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

Example letter for physician to provide to school

“My patient_______________________ has been diagnosed with Kabuki Syndrome a a rare genetic condition. It is characterized by distinctive facial features, growth and developmental delays, and speech and language difficulty. This condition necessitates an Individualized Education Program (IEP) or 504. Medical complications with Kabuki Syndrome can include possible seizures, feeding difficulty, heart defects, and increased risk for infection. Because of these, __________ needs the following accommodations:”

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.

Thank you so much to Susanne and family for sharing their story! And thank you to the team at the Institute on Disability at UNH for assisting in collecting these stories.