Kabuki syndrome (KS)
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

Physical characteristics and/or symptoms

Note: not all people with Kabuki syndrome will have all of these features.

Physical features

- Typical facial features in KS
  - Larger appearing eyes due to elongated palpebral fissures
  - Eversion of the lateral third of the lower eyelid
  - Arched and broad eyebrows
  - Large, prominent or cupped ears
- Minor skeletal differences
  - Spine
    - Butterfly vertebrae
    - Scoliosis
  - Short fingers and toes
  - Curved 5th finger
- Persistence of fetal fingertip pads
- Mild to moderate intellectual disability
- Growth deficiency
  - Small head size which may/may not accompany short stature.

Additional findings

- Congenital heart defects
- Genitourinary differences
  - Abnormal kidneys or urinary tract
- Cleft lip and/or palate
- Gastrointestinal problems
  - Anal atresia
  - Reflux

    - Diarrhea
    - Constipation
    - Feeding difficulties
- Eye findings
    - Eye lid droops
    - Crossed eyes
    - Coloboma
    - 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
- 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
    - Usually controlled with medicine
- Endocrinology abnormalities
    - Premature breast development
    - Hyperinsulinism in neonates and infants with persistent hypoglycemia
- Small mouth, small jaw
- Hearing loss (conductive) and ear pits
- Low muscle tone
- Loose joints
- Autism or autistic-like features

Recommended Routine Surveillance

- Assessment of feeding/eating, digestive problems
- Surveillance for cardiac abnormalities
- Monitor hearing and vision annually
- Monitor for seizures

- Brain imaging is recommend in those with headaches, ocular disturbances, lower cranial nerve signs, cerebellar ataxia, spasticity, or seizures to evaluate for a Chiari 1 malformation or for other brain malformations.
- Echocardiogram to evaluate for cardiac defect

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.
- If seizures are present, the following seizure action plan may be useful: https://www.epilepsy.com/learn/managing-your-epilepsy/seizure-action-plans

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
  - Congenital heart defects
- Craniofacial Multidisciplinary Team
  - Plastic surgeon to treat cleft lip/palate
  - Speech pathologist
  - Dentist/Orthodontist
  - Other team members
- Dentist
  - Dental anomalies
- Developmental evaluation:
  - Speech therapy
  - Physical therapy
  - Occupational therapy
  - Autism spectrum disorder /autistic signs
- ENT
  - Chronic otitis media
  - Hearing loss
- Endocrinologist
  - Premature thelarche
- Gastroenterologist
  - Feeding problems
  - Diarrhea
  - Constipation
- Geneticist / Genetic Counselor:

    - Diagnosis
    - Coordination of care
    - Genetic risk for family
    - Clinical trials
- Genitourinary
    - Renal anomalies
    - Urinary tract anomalies
- Immunologist
    - immune dysfunction
- Neurology
    - Monitor seizures
- Ophthalmologist
    - Strabismus
    - Ptosis
- Orthopedists
    - Skeletal anomalies
    - Joint laxity
    - Joint dislocations

Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient_____ has been diagnosed Kabuki syndrome. Kabuki syndrome is characterized by typical facial features, minor skeletal anomalies, postnatal growth deficiency, and developmental delays. Medical complications with Kabuki syndrome include management of seizures, increased susceptibility to infections and autoimmune disorders, endocrinology abnormalities, feeding problems, and hearing loss. Because of these, _____ needs the following accommodations.

Related Information:
• What does it mean if a disorder seems to run in my family? • What is the prognosis of a genetic condition? • Genetic and Rare Diseases Information Center
• Sample letters for parents to send to schools to explain the condition:
https://lpa.memberclicks.net/assets/school%20letters%202016.pdf

Seven Important Aspects of School Life

"Kabuki Syndrome at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

GeneReviews: Kabuki Syndrome
http://www.ncbi.nlm.nih.gov/books/NBK62111/

Kabuki Syndrome Network
www.kabukisyndrome.com

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/kabuki-syndrome