

Kabuki Syndrome: A Summary of Select Tables from GeneReviews



The tables below are selections from GeneReviews: Kabuki syndrome. They are intended to be provided to healthcare professionals for review and implementation, where appropriate. Additional diagnostic and management considerations are available at the link below.



The Kabuki Syndrome Foundation (KSF) increases our community's access to resources; we do not provide medical advice. Access the full GeneReviews article by scanning the QR code or visiting: www.ncbi.nlm.nih.gov/books/NBK62111/

Table 5. Recommended Surveillance for Individuals with Kabuki Syndrome

System/Concern	Evaluation	Frequency
Growth	Measurement of at least height & weight ¹	At each appointment
Ophthalmologic	Ophthalmology or optometry to assess vision	At least annually
Hearing	Hearing assessment	
Musculoskeletal	Clinical eval for scoliosis	At each appointment until skeletal maturity
Endocrinologic	Thyroid function tests	Every 2-3 yrs
Immunologic	Assessment of complete blood count	
Miscellaneous/ Other	Monitor developmental progress & educational needs	At each visit during childhood & adolescence

1. Adolescents and adults may develop obesity.

Table 3. Evaluations Following Initial Diagnosis

To establish the extent of disease and the needs of an individual diagnosed with KS, the following evaluations are recommended if they have not already been completed:

System/Concern	Evaluation	Comment
Growth	Measurement of height, weight, & head circumference	FTT is a common sequela of feeding difficulties.
Ophthalmologic	Ophthalmology eval	For assessment of strabismus, refractive error, ptosis, & corneal abnormalities
Hearing	Baseline audiology eval	To assess for conductive &/or sensorineural hearing loss
Mouth	Directed evaluation of the palate for palatal anomalies	Consider referral to a craniofacial specialist if palatal anomalies are suspected
	Consider dental eval for those age >3 yrs	
Cardiac	Echocardiogram w/visualization of the aortic arch	To assess for congenital heart defects incl coarctation of the aorta
	Consider EKG.	If arrhythmia is suspected
Respiratory	Consider chest radiographs to assess for diaphragmatic eventration.	In those w/respiratory issues, chronic cough, or recurrent pneumonia
Gastrointestinal/ Feeding	Assess nutritional status, feeding, GERD	<ul style="list-style-type: none"> Consider assessment by feeding team &/or VFSS for those w/ suspected dysphagia Infants may have FTT; adolescents & adults may have obesity
Genitourinary	Baseline renal ultrasound	To evaluate for renal anomalies & hydronephrosis
	Physical exam for hypospadias &/or cryptorchidism in males	

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Continued: Table 3. Evaluations Following Initial Diagnosis

System/Concern	Evaluation	Comment
Musculoskeletal	Consider radiographs of the spine in those w/scoliosis	To assess for vertebral anomalies
Endocrinologic	Assess for hyperinsulinism ¹	In neonates & infants w/persistent hypoglycemia
	Assess for hypothyroidism & growth hormone deficiency ²	In those w/abnormal growth velocity
Immunologic	T cell count, T cell subsets, & serum immunoglobulin levels at time of diagnosis or at age 1 yr (whichever is later)	Refer to immunologist if: <ul style="list-style-type: none"> • Levels are abnormal; or • Patient has history of recurrent infections
Neurologic	EEG	In those w/suspected seizures
	Head MRI	To evaluate for: <ul style="list-style-type: none"> • Structural brain malformations in those w/ seizures • Chiari I malformations in those w/ suggestive symptoms³
Psychiatric/ Behavioral	Neuropsychiatric eval	Screen persons age >12 mos for behavior concerns incl sleep disturbances, ADHD, anxiety, &/or traits suggestive of ASD
Miscellaneous/ Other	Developmental assessment	Evaluate motor, speech/language, general cognitive, & vocational skills
	Consultation w/clinical geneticist &/or genetic counselor	

ADHD = attention-deficit/hyperactivity disorder; ASD = autism spectrum disorder; FTT = failure to thrive; GERD = gastroesophageal reflux disease; VFSS = videofluoroscopic swallowing study

1. This may include collection of a "critical sample," such as obtaining plasma levels of insulin, free fatty acids, beta-hydroxybutyrate, and glycemic response to glucagon during a period of low plasma glucose [Yap et al 2019].
2. Thyroid function tests may include free T4 and TSH levels. Assessment for growth hormone deficiency can be challenging and is best directed by an endocrinologist. Tests may include measurement of insulin-like growth factor 1 (IGF-1) and IGF binding protein 3, in addition to consideration of a growth hormone stimulation test using either arginine or clonidine [Schott et al 2016b].
3. Symptoms may include headaches, ocular disturbances, otoneurologic disturbances, lower cranial nerve signs, cerebellar ataxia, spasticity, or seizures.

Thank you to the authors of GeneReviews: Kabuki Syndrome: Adam MP, Hudgins L, Hannibal M. Kabuki Syndrome. 2011 Sep 1 [Updated 2022 Sep 15]. In: Adam MP, Mirzaa GM, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK62111/>

Questions for your physician? You can write them here:

More resources for people with Kabuki syndrome and caregivers are available at www.KabukiSyndromeFoundation.org.

