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Case Report

Kabuki Syndrome and Anesthetic Challenges in Pediatric Upper Gastrointestinal Endoscopy: A Focused Narrative Review with a Clinical Illustration

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Abstract

***Background*:** Kabuki syndrome is a rare developmental disorder characterized by distinctive craniofacial features and variable multisystem involvement. Its anesthetic implications extend beyond potential difficult intubation to respiratory vulnerability, swallowing dysfunction, cardiovascular abnormalities, and neurological comorbidities. These concerns are particularly relevant during upper gastrointestinal endoscopy, when the endoscopist and anesthesiologist share access to the upper airway. ***Clinical illustration*:** A boy with a reported pathogenic KDM6A variant underwent esophagogastroduodenoscopy for investigation of dysphagia. His phenotype included a high arched palate, axial hypotonia, developmental delay, and a history of seizures treated with valproate. Previous echocardiography showed no structural cardiac abnormality. The clinical presentation and procedural photographs provide an opportunity to revisit the anesthetic implications of this uncommon syndrome. ***Discussion*:** This narrative review addresses airway assessment, aspiration risk, respiratory depression, neuromuscular recovery, associated organ disease, and preparation for rescue interventions. Published anesthetic experiences demonstrate heterogeneous clinical presentations and support individualized planning. Concerns about malignant hyperthermia and latex allergy should be interpreted according to the strength of the available evidence and the individual history. ***Conclusion*:** Recognition of Kabuki syndrome should prompt a phenotype based anesthetic assessment even for brief diagnostic procedures. During upper gastrointestinal endoscopy, coordinated airway management and attention to dysphagia, hypotonia, and recovery are central to perioperative planning.

Keywords: Kabuki syndrome, KDM6A, pediatric anesthesia, upper gastrointestinal endoscopy, dysphagia, airway management.

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INTRODUCTION

Kabuki syndrome is an uncommon developmental condition, with an estimated occurrence of approximately one in 32,000 newborns [1]. Its rarity means that an anesthesiologist may encounter an affected patient only occasionally, despite the need for repeated investigations or surgery. Variants in KMT2D or KDM6A affect chromatin regulation and contribute to a variable phenotype [1,2]. The anesthetic challenge arises from the combination of anatomical and functional abnormalities rather than from a uniform response to anesthetic drugs.

A child referred for a short endoscopic examination may have craniofacial abnormalities, hypotonia, feeding problems, or organ involvement that warrants additional preparation. Using a boy undergoing esophagogastroduodenoscopy (EGD) for dysphagia as a clinical illustration, we review the syndrome and the practical challenges it presents to the anesthesia team.

CLINICAL ILLUSTRATION

A boy with Kabuki syndrome underwent EGD for investigation of dysphagia. The diagnosis had been established in the context of developmental delay and a characteristic clinical phenotype. Specialist reports documented a pathogenic variant in KDM6A. Conventional cytogenetic analysis showed a 46, XY karyotype.

His neonatal history included respiratory distress requiring an eight-day admission to neonatal intensive care, with favorable subsequent progress. Feeding difficulties and hypotonia were reported during infancy. The phenotype included long eyelashes, large low set ears, a high arched palate, prominent fingertip pads, bilateral single transverse palmar creases, strabismus, and left testicular ectopia. Axial hypotonia and developmental delay were prominent features of the neurological assessment.

The medical history also included seizures associated with episodes of vomiting. A pediatric report documented treatment with valproate and multidisciplinary rehabilitation. Brain magnetic resonance imaging showed no parenchymal lesion. Echocardiography performed in infancy was reported as normal, with preserved left ventricular systolic function; abdominal ultrasonography was also described as unremarkable.

EGD was performed to investigate dysphagia. The intraoral image illustrates the high arched palate (Figure 1), and the procedural image shows oral endoscopy with nasal oxygen (Figure 2). Esophagogastroduodenoscopy was performed under deep sedation with propofol target-controlled infusion using the Schnider pharmacokinetic model, titrated to a Modified Observer's Assessment of Alertness/Sedation (MOAA/S) score of 1–2. Supplemental oxygen was delivered through nasal cannulae incorporating capnographic sampling.

Practical Implications for Upper Gastrointestinal Endoscopy

Clinical Figures



Figure 1: Intraoral view of the child with Kabuki syndrome, illustrating the palatal morphology corresponding to the high arched palate described in the clinical examination. This feature is relevant to preanesthetic airway assessment



Figure 2: Esophagogastroduodenoscopy performed for investigation of dysphagia. The photograph shows the oral endoscope and nasal oxygen interface, illustrating the shared upper airway during the procedure

Recognizing the syndrome

The international diagnostic consensus emphasizes infantile hypotonia and developmental impairment in conjunction with characteristic dysmorphism and/or a pathogenic molecular finding [2]. Typical manifestations include long palpebral fissures, characteristic eyebrows and ears, persistent fetal fingertip pads, growth impairment, and skeletal abnormalities. A high arched or cleft palate and dental anomalies may further influence airway assessment. KDM6A associated disease is X linked, whereas KMT2D associated disease generally follows an autosomal dominant pattern [1, 2].

Recognition has immediate clinical value: apparently unrelated findings such as feeding difficulty, hypotonia, and facial dysmorphism may belong to the same multisystem disorder. The genetic diagnosis directs attention to possible associated disease; the actual examination and medical history determine its relevance to anesthesia.

Concerning anesthetic challenges, airway anatomy and procedural access, a high arched palate, abnormal dentition, malocclusion, or other craniofacial abnormalities may complicate airway instrumentation. OrphanAnesthesia recommends careful airway and cardiopulmonary assessment and preparation for possible difficult intubation [3]. In the illustrative child, the palatal feature is clinically relevant without implying that difficult intubation is inevitable. Atalay *et al.,* reported straightforward tracheal intubation despite craniofacial abnormalities in a patient undergoing scoliosis surgery [4].

EGD adds a procedural constraint: the endoscope occupies the mouth and pharynx while the anesthesiologist must preserve oxygenation and ventilation. Pediatric endoscopy guidance identifies craniofacial abnormalities and cardiopulmonary disease as factors associated with increased procedural risk [5]. For a child with Kabuki syndrome, an explicit agreement about interruption and scope withdrawal is a practical application of these principles. Suction, age-appropriate ventilation equipment, and an alternative airway strategy should be immediately accessible.

Feeding difficulty in Kabuki syndrome may reflect impaired oromotor coordination, hypotonia, palatal abnormalities, or swallowing dysfunction; gastroesophageal reflux also occurs [6]. Dysphagia therefore provides both an indication for investigation and a reason to assess airway protection. The preanesthetic history should characterize choking, coughing during meals, regurgitation, previous aspiration, and feeding support. These features inform the assessment; dysphagia alone does not establish that aspiration has occurred.

The choice between an uninstrumented airway and tracheal protection should account for swallowing symptoms, respiratory status, procedural requirements, and anticipated rescue difficulty. This is an individualized clinical judgment rather than a syndrome specific rule. NASPGHAN guidance supports risk stratification before pediatric endoscopy [5]. A short-anticipated procedure does not remove the need to plan for respiratory compromise.

Hypotonia may coexist with obstructive sleep apnea, ineffective cough, or airway abnormalities. Elmitwalli *et al.,* emphasize these concerns and the potential need for postoperative respiratory monitoring [7]. Assessment should include baseline breathing, snoring, respiratory infections, and the child's functional status. The goals are to identify vulnerability and to plan observation appropriate to the individual.

When neuromuscular blocking drugs are used, recovery requires objective assessment. General ASA guidance recommends quantitative monitoring at the adductor pollicis and confirmation of a train of four ratio of at least 0.9 before extubation [8]. Drug and reversal choices must be appropriate for the child's age and clinical circumstances. Kabuki syndrome should not be assumed to confer a predictable degree of sensitivity to every neuromuscular blocking agent.

Cardiac malformations belong to the Kabuki phenotype and can materially alter hemodynamic goals [6]. A prior normal echocardiogram is reassuring, but assessment should still consider current symptoms and the available cardiology history. The 2026 report of dental anesthesia in a child with Kabuki syndrome and Fontan circulation illustrates how associated cardiac physiology can dominate anesthetic planning [9]. A technique selected for that physiology cannot be generalized to all affected children.

Seizures and developmental impairment affect perioperative preparation. Antiseizure treatment should be reviewed and unnecessary interruptions avoided; communication with caregivers helps establish baseline behavior and neurological function [7]. In the illustrative case, the history of seizures and valproate use complements the airway assessment by identifying another aspect of the syndrome relevant to procedural care.

Renal abnormalities, endocrine disturbances, and immune dysfunction may accompany Kabuki syndrome [6]. Their presence and clinical consequences should be established from the individual history and targeted investigations. Joint laxity also warrants attention during positioning, particularly around the cervical spine [3]. The syndrome should prompt a focused search for these problems without attributing every described manifestation to every patient.

Malignant hyperthermia is discussed as a concern in some anesthetic reports and older guidance [3,10]. The selected literature does not establish a universal susceptibility or quantify an increased incidence attributable to Kabuki syndrome. Hypotonia alone should not be presented as proof of susceptibility. Personal and family anesthetic histories remain important. Conversely, uneventful use of sevoflurane in a published case cannot establish the absence of risk across the syndrome [9].

Teixeira *et al.,* reported confirmed latex allergy in a patient with Kabuki syndrome who subsequently underwent an uneventful procedure with latex avoidance [11]. This observation supports asking specifically about prior reactions and implementing appropriate avoidance when allergy is suspected or established. It does not demonstrate that all patients with Kabuki syndrome are latex allergic.

No single anesthetic technique is appropriate for every patient with Kabuki syndrome [3]. Çatalca *et al.,* described inadequate ventilation through a laryngeal mask followed by successful tracheal intubation [10], illustrating the need for an alternative airway strategy. During EGD, sedation depth and airway protection should be selected according to swallowing symptoms, current respiratory function, anticipated intervention, and rescue access. Nasal oxygen alone does not demonstrate adequate ventilation. Practical implications for upper gastrointestinal endoscopy apply the reviewed principles to the procedural setting, assess airway anatomy and swallowing symptoms together: palatal morphology and dysphagia address different components of respiratory risk; clarify the current neurological and cardiorespiratory baseline, including

antiseizure treatment and any history of respiratory obstruction; agree with the endoscopist on immediate scope withdrawal if airway access or ventilation is required and prepare an alternative airway strategy and base recovery observation on respiratory function and comorbidities rather than procedure duration alone.

CONCLUSION

Kabuki syndrome is rare, but its clinical features are directly relevant to everyday anesthetic decisions. The child undergoing EGD for dysphagia provides a reminder that an apparently limited diagnostic procedure can bring together anatomical airway concerns, swallowing dysfunction, and hypotonia. Recognizing the syndrome, assessing the individual phenotype, and preparing a coordinated respiratory rescue strategy are the central lessons. Published experience supports adaptable planning rather than a universal anesthetic protocol.

Published anesthetic experience

Table 1: Selected reports illustrating the diversity of anesthetic concerns in Kabuki syndrome. These observations illustrate clinical decisions and do not provide syndrome specific complication rates

Report	Clinical setting	Relevant observation
Teixeira 2010 [11]	11 year old girl; soft tissue surgery.	Confirmed latex allergy; uneventful subsequent anesthesia with latex avoidance.
Atalay 2014 [4]	Revision surgery for scoliosis.	Cormack–Lehane grade II view and easy intubation despite craniofacial features; careful positioning.
Elmitwalli 2023 [7]	Infant; airway surgery and later hip surgery.	No reported mask ventilation or intubation difficulty; comorbid organ involvement informed care.
Çatalca 2024 [10]	6 year old boy; strabismus surgery.	Elevated inspiratory pressures and secretions after laryngeal mask placement; conversion to tracheal intubation.
Chizuwa 2026 [9]	10 year old girl; dental treatment; Fontan circulation.	Anesthesia included remimazolam, fentanyl, sevoflurane, and rocuronium; no perioperative complication reported.

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