

Anesthesia for a child with Kabuki Syndrome

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STR—Kabuki syndrome is a rare multiple malformation/mental retardation syndrome. The exact cause remains unknown and multiple possible genetic abnormalities have been reported. Drs Niikawa and Kuroki first described the disease in 10 unrelated children in 1981 (1). The name ‘Kabuki make-up syndrome’ is from the similarities of the faces of affected children with make-up from traditional Japanese (Kabuki) theater.

We report the case of a 5-year-old Hispanic female scheduled for dental fillings and auditory brainstem response test. She weighed 14 kg and was 37in tall which put her on the 1 percentile for weight, and <1% for height. Her postnatal growth retardation and physical characteristics such as long palpebral fissures, large prominent fissures with eversion of the lateral third of the lower eyelids, and large prominent malformed ears are classic for her diagnosis of Kabuki syndrome. Dental abnormalities and hearing loss are common (1).

Her mother reported a history of heart murmur, and as an infant she was diagnosed with left pulmonary artery narrowing and a patent foramen ovale. However, her current echocardiogram was reported as normal. As an infant she had frequent pneumonia from reflux and aspiration and spent her first 2 months of life ‘in and out’ of the Neonatal intensive care unit. At presentation for preoperative evaluation, she had no heart murmur. Lung sounds were clear bilaterally. Mallampati score was II and she was very cooperative.

Past history revealed that she had undergone anesthesia and surgery in our institution without incident for several bilateral ear tubes, a Nissen fundoplication, cleft palate repair, and gastrostomy tube using a combination of sevoflurane, propofol, and fentanyl.

She was premedicated with 0.5 mg/kg⁻¹ midazolam elixir and 10 mg/kg⁻¹ ibuprofen elixir. Anesthesia was induced using 8% sevoflurane in a 50% N₂O, 50% O₂ mixture. A 22-gauge peripheral i.v. was started.

A grade I laryngoscopic view was obtained using a #2 Macintosh blade and the trachea was intubated with a 5.0 uncuffed orotracheal tube. Anesthesia was maintained with sevoflurane 1.8% in 50% N₂O, 50% O₂ mixture with spontaneous breathing throughout. At the completion of surgery she was extubated and the postoperative course was uncomplicated.

There are 300 cases of Kabuki syndrome reported. This syndrome was first described in Japanese children but occurs in many other ethnic groups (1). At present there is no test for the syndrome and the diagnosis is made from the physical characteristics. Most are affected by a new mutation. Based on the rare instance of inheritance the disorder is most likely autosomal dominant (2).

Characteristic facial features include long palpebral fissures with eversion of the outer third, ptosis, arched eyebrows, prominent eyelashes, misshapen prominent ears and short nasal septum (2). These children have postnatal growth deficiency, which is noticeable in the first year, skeletal abnormalities, mild to moderate intellectual disability, and short stature. Although mental retardation is common, most have good social skills and are at ease with strangers (1). Approximately one-third has seizures, and can be of any type (1).

Abnormalities that could affect anesthesia care include micrognathia, cleft and high arched palate and could lead to possible difficult airway; no incidence has yet been reported. Pulmonary function is usually normal, but obstructive sleep apnea has been reported in several patients in their teens (1), likely because of hypotonia and that children with Kabuki syndrome tend to become obese in later childhood (2). Scoliosis is present in 35% (1) which can affect respiratory function (3).

Approximately half have cardiac abnormalities, which are usually diagnosed early in life (2) and include coarctation of the aorta, bicuspid aortic valve, mitral valve prolapse, ventricular septal defect, valvular stenosis, tetralogy of Fallot, single ventricle with common atrium, double outlet right ventricle, or transposition of the great vessels (2). Renal abnormalities occur in 25% of patients with Kabuki syndrome.

Muscular hypotonia is also a common feature and concerns have been raised for neuromuscular blockade and malignant hyperthermia but muscle biopsies have been normal (1). Remifentanyl has been reported as an alternative to inhalation anesthesia without complication (4). Others have reported larger than usual requirements for nondepolarizing muscle relaxants, possibly because of concomitant anticonvulsant therapy. No post-anesthesia respiratory problems were reported in that case (5).

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