Case Report

Anesthesia management in Kabuki make-up syndrome

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ABSTRACT

Kabuki make-up syndrome (KMS) is a rare condition with a number of characteristic congenital abnormalities. The syndrome is characterized by peculiar facial appearance (resembling the make-up of actors in Kabuki, the traditional Japanese theater), skeletal anomalies, dermatoglyphic abnormalities, postnatal growth deficiency, and mental retardation. These are rare reports of central nervous system dysfunctions, other than mental retardation, and no previously described congenital talipes calcaneo-valgus in this syndrome. We report the case of a 22-month-old girl having Kabuki make-up. At presentation, she had an adenoid hypertrophy and a history of recurrent otitis media. She had also delay in motor development, and a postnatal growth deficiency. The variable phenotypic expression is a well-known characteristic of the syndrome. For that reason, we should perform careful morphologic examination in every patient and their parents, and use flexotype laryngoscope (Heine, Germany) to visualize vocal cord in case of difficult intubation. At preoperative examination, as clinicians, we must be careful regarding patient morphology. Congenital heart defects and epilepsy are important for anesthesia management in KMS.

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Kabuki make-up syndrome (KMS) was first described in 1981 by Niikawa and Kuroki et al.[1,2] The 5 cardinal manifestations of the syndrome are, according to Niikawa et al,[1] a peculiar face characterized by eversion of the lower lateral eyelid, arched eyebrows with sparse or dispersed lateral one third, a depressed nasal tip and prominent ears; skeletal anomalies including brachydactyly V, and a deformed spinal column; dermatoglyphic abnormalities including increased digital ulnar loop and hypothenar loop patterns, absence of the digital triradius, or both, and presence of fingertip pads; mild to moderate mental retardation; and postnatal growth deficiency.[2] In addition, previous studies observed many other inconsistent anomalies such as a precocious puberty, congenital heart defects, palatal abnormalities, ectodermal abnormalities, neurologic dysfunctions, cerebellar, and brainstem atrophy, and recurrent dislocation of the patella.[3-5] The cause of KMS, which is seen sporadic, is presently unknown. However, proposed X-linked and autosomal dominant inheritance based on the presence of minor manifestations in a few of parents. In most cases, Say et al,[6] reported conductive hearing loss as sensorineural and mixed type, it is associated with recurrent otitis media. As pointed out, recognition of the phenotypic spectrum, natural history, and prognosis of a genetic disorder are critical to proper patient care.

Case Report. A 22-month-old girl, who is the first baby of healthy family, 73 cm height (under 3rd percentile), 7.65 kg weight (under 3rd percentile), and 43 cm head circumference (97% percentile). She was born at term, after uneventful pregnancy, by sectio ceasarian, and weighed 2.2 kg, and due to low birth weight (LBW), we admitted

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Figure 1 - Patient with Kabuki make-up syndrome showing arched eyebrows, long eyelashes and asymmetric prominent ears.

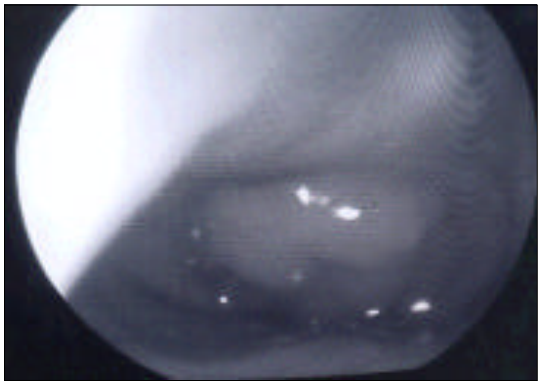

Figure 2 - Endoscopic view of adenoid hypertrophy.

her to the Special Care Baby Unit. We diagnosed her as having Kabuki make-up syndrome. The diagnosed was made by geneticists based on the presence of facial characteristics and clinical genetic reviews. At birth she was noted to have an asymmetric prominent ear, arched eyebrows, long eyelash, epicanthus, phalanx deformity of hands, broad thumbs, a mild to moderate delay in motor development, and a postnatal growth deficiency (**Figure 1**). She was also noted to have anal stenosis and a ventricular septal defect. At 7th day, she underwent surgery for anal stenosis. We performed a complete ear–nose–throat examination that was supported with nasal and nasopharyngeal endoscopy for assessment of nasal patency and adenoid size. She was premedicated with midazolam 0.15 mg kg^{-1} and atropine 10 µg kg^{-1} 30 minutes before the induction of anesthesia intravenously (IV). We induced anesthesia with thiopental 5 mg kg^{-1}, fentanyl 2 µg kg^{-1}, and rocuronium bromur 0.5 mg kg^{-1} 100% in oxygen. We used a 70 degree curve flexotype laryngoscope blade, (Heine, Germany) so as to visualize the vocal cords in case of difficult intubation. After tracheal intubation, we set the fresh gas-flow rate to 4.4 L minute. We adjusted anesthesia maintenance to maintain air 50% in oxygen, IV fentanyl 1-2 µg kg^{-1}, and rocuronium bromur 0.2 mg kg^{-1} repeatedly. Volatile anesthetic concentration was adjusted to maintain for sevoflurane 1 minimum alveolar concentration (MAC) with systolic blood pressure within ± 20% of baseline. We controlled ventilation with a tidal volume of 7-8 $mLkg^{-1}$ and adjusted the respiratory rate to maintain with end-tidal carbon dioxide ($EtCO_2$) value between 35-45 mm Hg. The anesthetic machine used was datex-ohmeda ADU® Anesthesia System (Datex-Ohmeda, S/5, Helsinki, Finland). She was monitored by electrocardiography, noninvasive blood pressure, peripheral oxygen saturation (SpO_2), and end-tidal CO_2 measurement. During anesthesia, the end-tidal CO_2 concentration inspired and end-tidal anesthetic concentrations, were monitored by mass spectrometry (Datex Ohmeda, ADU, S/5, Helsinki, Finland). She underwent an adenoidectomy operation under general anesthesia with curettage and cold dissection method due to obstructive adenotonsillar hypertrophy. She experienced recurrent otitis media necessitating placement of bilateral ventilation tubes. These were placed initially at the age of 22 months. We collected the data when she underwent surgery for ventilation tubes and adenoidectomy at 22-months-old (**Figure 2**).

Discussion. In recent years, a growing number of cases of KMS have been reported, and most reported cases are Japanese patients. The variable phenotypic expression is a well-known characteristic of the syndrome. For that reason; we should perform careful morphologic examination in every patient and their parents. Kristiane et al,[7] observed congenital heart defects along with many other inconsistent anomalies. Some investigators suggest epilepsy to be one of the cardinal features of KMS.[8] In a recent review on this topic, Schrander et al,[9] reported epilepsy in 14% of the Japanese patients and in 19% of the non-Japanese, but electroclinical data were not analyzed. In these patients, we prefer sevoflurane or midazolam and opioid drugs, as anesthetic drugs, such as propofol, may cause an epileptic episode. We should use a flexotype laryngoscope blade to visualize the vocal cord in case of difficult intubation. At preoperative examination, as clinicians, we must be careful regarding patient morphology. Congenital heart defects and epilepsy are important for anesthesia management in KMS.

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