

## Acknowledgement

The work was presented at the 12th World Congress of Anaesthesiologists, 4–9 June 2000, Montreal, Quebec, Canada.

## References

1. Weinger MB, Englund CE. Ergonomic and human factors affecting anesthetic vigilance and monitoring performance in the operating room environment. *Anesthesiology* 1990; 73: 995–1021.
2. Broadbent DE, Cooper PE, FitzGerald P, Parkes KR. The Cognitive Failures Questionnaire (CFQ) and its correlates. *Br J Clin Psychol* 1982; 21: 1–16.
3. Rasmussen K, Jeppesen HJ, Sabroe S. Psychometric tests for assessment of brain function after solvent exposure. *Am J Ind Med* 1993; 24: 553–565.
4. Araki S, Murata K. Determination of evoked potentials in occupational and environmental medicine: a review. *Environ Res* 1993; 63: 133–147.
5. Stollery BT, Broadbent DE, Lee WR, Keen RI, Healy TEJ, Beatty P. Mood and cognitive functions in anaesthesiologists working in actively scavenged operating theatres. *Br J Anaesth* 1988; 61: 446–455.
6. Lucchini R, Belotti L, Casitto MG, et al. Neurobehavioral functions in operating theatre personnel: a multicenter study. *Med Lav* 1997; 88: 396–405.
7. Mahoney AM, Dalby JT, King MC. Cognitive failures and stress. *Psychol Rep* 1998; 82: 1432–1434.

## Anaesthetic management in a case of Kabuki syndrome

### EDITOR:

The Kabuki syndrome is a rare congenital disease characterized by distinct craniofacial dysmorphias; skeletal, skeletal muscle and dermatoglyphic abnormalities; cardiovascular, visceral and urogenital anomalies; postnatal growth deficiency; mild to moderate mental retardation; neurological dysfunction and susceptibility to infections [1]. Although patients afflicted with this syndrome require operative correction of the multiple anomalies, we have only found one case report concerning to the anaesthetic management in this disease [2].

Our patient was a 14-month-old female, 6.600 kg, diagnosed with Kabuki syndrome and scheduled for cleft palate repair. She had microcephaly, right microphthalmia, convergent strabismus, bilateral pupillary coloboma, long palpebral fissures with eversion of the lateral third of the lower eyelids, large prominent earlobes with low implantation, generalized hypotonia, megacisterna subaracnoidea and bilateral renal hypoplasia with normal renal function. The patient was premedicated with intranasal midazolam 0.3 mg kg<sup>-1</sup>. Due to difficulty in establishing venous access, we opted for an inhalation induction with sevoflurane 5% in pure O<sub>2</sub> and we were then able to cannulate a peripheral vein. Spontaneous breathing was maintained. Atropine 15 µg kg<sup>-1</sup> and remifentanyl 1 µg kg<sup>-1</sup> were given, and laryngoscopy showed a Grade III Cormack and Lehane view of the larynx. The trachea was

intubated with a 4 mm reinforced cuffed tube with the help of an introducer to guide and stiffen it. Anaesthesia was maintained with sevoflurane 1.5% in a mixture of 50% N<sub>2</sub>O in O<sub>2</sub> and remifentanyl 0.1–0.2 µg kg<sup>-1</sup> min<sup>-1</sup>. Once the operation was finished, we stopped the remifentanyl infusion and before extubation, we administered morphine 0.4 mg, ondansetron 0.6 mg and paracetamol 100 mg. The intraoperative and postoperative course was uneventful (Fig. 1).

Niikawa and Kuroki first described the Kabuki syndrome [3,4] in 1980. It was originally known as Kabuki make-up syndrome because of the resemblance of the facial features – Kabuki syndrome – and the make-up used in traditional Japanese theatre. The



Figure 1.  
Our patient after the cleft palate repair. (With permission of parents.)

Correspondence to: Ana Casado Merodio, López de Luna n° 16, 50009 Zaragoza, Spain. E-mail: aicasado@comz.org; Tel: +34 976567931

Accepted for publication February 2003 EJA 1415

aetiology of the Kabuki syndrome is still unclear, but most cases are sporadic and compatible with an autosomal dominant mutation with variable expressivity. However, a small number of patients have shown chromosomal abnormalities [5]. Our patient had a 46-XX karyotype, without any chromosomal abnormality. There were not family antecedents of the Kabuki syndrome, but the child's mother previously underwent a termination of pregnancy because of a fetal polymalformative syndrome.

Children with Kabuki syndrome have similar facial features, most notably lower palpebral eversion, long palpebral fissures, arched eyebrows, long eyelashes, blue sclerae, depressed nasal tip, cleft lip/palate, arched palate, dysmorphic ears and abnormal dentition. Joint laxity is common and skeletal anomalies include brachydactyly, clinodactyly and deformed vertebrae or ribs. Van Haelst and colleagues [6] reported two patients with stenosis of the central airways (one with local stenosis of the right upper lobe bronchus and the other with severe bronchomalacia and an abnormal right bronchial tree). Hence, the anaesthesiologist needs to be especially prepared for difficulty with the airway. We had a laryngeal mask ready for any possibility of difficult intubation but, although the vocal cords were not visible at laryngoscopy, the trachea was intubated with the help of a long introducer to stiffen the tube. The laryngeal mask seems a good alternative in case of problems; other methods include fiberoptic laryngeal intubation (although it is limited to tracheal tubes of at least 4.5–5.0 mm internal diameter), tracheostomy and retrograde intubation – all these carry risks and require considerable skill.

The risk of anaesthesia is also increased in these patients because of cardiovascular, urogenital and neurological problems, and abnormalities of the skeletal muscles. Children with Kabuki syndrome often have hypotonia, seizures and microcephaly. Digilio and colleagues [7] presented the results of cardiac evaluations of 60 patients diagnosed with Kabuki syndrome at their institution. Cardiac evaluation included chest radiography, electrocardiogram and two-dimensional and colour Doppler echocardiography. Thirty-five of the patients (58%) had congenital heart defects. The most commonly observed defects were coarctation of the aorta (23%), atrial septal defect (20%) and ventricular septal defect (17%). Our patient had been studied previously and a cranial CAT scan, an electroencephalogram, an abdominal ultrasound and a Doppler echocardiography were available. Due to the existing hypotonia and the possible difficult airway, we did not use any neuromuscular blocking

drugs and we made use of remifentanyl. With this drug, we obtained good conditions for tracheal intubation and we did not need a high dose to maintain haemodynamic stability and optimal conditions during surgery. Nevertheless, it is important to bear in mind the possible appearance of rigidity after the administration of remifentanyl. In a non-relaxed patient, it is essential to inject the drug slowly. The bolus,  $1 \mu\text{g kg}^{-1}$ , should be administered intravenously (i.v.) over 30 s; in case of difficulty to ventilate the lungs a muscle relaxant can be given.

Anaesthesiologists should be aware of a possible difficult tracheal intubation and multiples associated anomalies when dealing with the anaesthetic management of a patient with Kabuki syndrome. We believe, on the basis of this single case, that remifentanyl – in view of its pharmacokinetic and pharmacodynamic features (small distribution volume, rapid metabolic clearance and short peak effect time) – may be suitable for use in these patients where the perioperative management can be complicated.

A. I. Casado, J. Ruiz, J. Oro, C. Martínez,  
I. Fernández, P. Oliva  
Department of Anaesthesiology  
Miguel Servet University Hospital  
Zaragoza, Spain

## References

1. Wessels MW, Brooks AS, Hoogeboom J, Niermeijer MF, Willems PJ. Kabuki syndrome: a review study of three hundred patients. *Clin Dysmorphol* 2002; 11: 95–102.
2. Wappler E, Standl T. Special anesthesiological aspects in patients with Niikawa-Kuroki ('Kabuki make-up') syndrome. *Anesthesiol Intensivmed Notfallmed Schmerzther* 1997; 32: 197–200.
3. Niikawa N, Matsuura N, Fukushima Y, et al. Kabuki make-up syndrome: a syndrome of mental retardation, unusual faces, large and protruding ears, and postnatal growth deficiency. *J Pediatr* 1981; 99: 565–569.
4. Kuroki Y, Suzuki Y, Chyo H, et al. A new malformation syndrome of long palpebral fissure, large ears, depressed nasal tip, and skeletal anomalies associated with postnatal dwarfism and mental retardation. *J Pediatr* 1981; 99: 570–573.
5. Online Mendelian Inheritance in Man, OMIM®. John Hopkins University, Baltimore, Md. MIM Number 147920; 3/3/96; URL: <http://www.ncbi.nlm.nih.gov/Omim/>
6. Van Haelst MM, Brooks AS, Hoogeboom J, et al. Unexpected life-threatening complications in Kabuki syndrome. *Am J Med Genet* 2000; 94: 170–173.
7. Digilio MC, Marino B, Toscano A, Giannotti A, Dallapiccola B. Congenital heart defects in Kabuki syndrome. *Am J Med Genet* 2001; 100: 269–274.

Copyright of European Journal of Anaesthesiology is the property of Greenwich Medical Media Ltd. and its content may not be copied or emailed to multiple sites or posted to a listserv without the copyright holder's express written permission. However, users may print, download, or email articles for individual use.