Kabuki syndrome is a rare multiple congenital anomalies/mental retardation (MCA/MR) syndrome that presents with craniofacial anomalies, mental retardation, skeletal, cardiovascular and renal abnormalities. Given this constellation of clinical findings that can affect the conduct of anesthesia, we report the peri-operative management of a child with Kabuki syndrome and discuss the potential problems in anesthesia.

Case Report: 16 month-old male developed the new onset of fevers and status epilepticus and was admitted to the intensive care unit for his seizure disorder and aspiration pneumonia. An MRI revealed a 2x2 cm mass at left temporal lobe and a left temporal craniotomy was scheduled after the patient’s pneumonia had resolved. His past medical history was significant for Kabuki syndrome, developmental delay, and seizure disorder with no congenital cardiac lesions. His seizures were treated with phenobarbital and he was seizure-free thereafter. Preoperative evaluation revealed BP 80/45, SpO2 99%, HR 120, regular rate and rhythm with no murmurs or rubs. An ECG confirmed the absence of any arrhythmias.

The patient underwent sevoflurane mask induction in the presence of his mother. Mask ventilation was easy. His vocal cords were easily visualized under direct laryngoscopy and his trachea was easily intubated with a 4.5 mm uncuffed tube. Anesthesia was maintained with isoflurane/N2O/O2 and fentanyl. His anesthetic course was remarkable for an increased requirement for non-depolarizing muscle relaxants. He maintained a stable cardiovascular course with no arrhythmias. The left temporal craniotomy revealed a 1.8 cm mass underneath an arachnoid cyst-like structure. The estimated blood loss was 50 mls. At the conclusion of the surgery, his trachea was extubated without any respiratory problems. He was transferred to ICU for postoperative observation. He did well except for one seizure episode without any sequelae and was discharged home 5 days later.

Discussion: Kabuki syndrome was first described independently by Drs. Niikawa and Kuroki in Japan in 1980. The majority of cases of Kabuki syndrome are sporadic and the sex ratio is nearly equal. The cause of this syndrome is currently unknown, although numerous cytogenetic abnormalities have been reported. The name “Kabuki” was selected because their face resemble the make-up of actors in Kabuki, traditional Japanese theater (arched eyebrows, long eyelashes, depressed nasal tip, and lower palpebral eversion). These dysmorphic features form the current basis for making the diagnosis. A review by Adam and colleagues report that failure to thrive and postnatal growth retardation occur during the first year of life. Obesity in adolescence follows. Hypotonia is frequently seen, although muscle biopsies in most cases have been normal. 10-39% of patients with Kabuki syndrome have seizures due to structural CNS lesions. Otitis media seems to be a frequent problem in these patients and half experience hearing loss. Cleft palate/lip have been described in 1/3 of patients. 40-50% of these patients have congenital cardiovascular anomalies; juxtaductal coarctation of the aorta, VSD, and ASD. Respiratory abnormalities are relatively uncommon, although obstructive sleep apnea may be seen with tonsil hypertrophy. Skeletal abnormalities can accompany, including short and incurved fifth digit, hip dislocation, vertebral abnormalities (butterfly vertebra, sagittal cleft, narrow intervertebral disc space, and scoliosis).

The anesthetic management of patients with Kabuki syndrome should focus on the various organ systems affected. Mental retardation leads to difficulty with communication with the patients and necessitates premedication for smooth induction. Given the dysmorphic facial features, airway management can be potential problem. Bueltler and colleagues listed airway complication as potential problem for sedation for the multiple imaging studies performed on these patients. A careful assessment of airway abnormalities and symptoms including sleep apnea should be performed. Cardiovascular status needs to be addressed in symptomatic patients. Echocardiogram may be helpful at the time of diagnosis if the patient’s physical examination warrants it. Renal function needs to be assessed and drugs that are eliminated via the renal
system may need to be avoided. Musculoskeletal problems such as hypotonia are frequently seen, but thus far there are no reports of malignant hyperthermia or muscular dystrophies associated with Kabuki syndrome. Most patients are on chronic anticonvulsant drug therapy and may demonstrate altered drug metabolism such as the increase requirements for non-depolarizing muscle relaxants.

References: