Anesthetic Management and Concerns of a Child with Marshall-Smith Syndrome

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Case Report: This 17-year-old patient with a traumatic rupture of the globe had Marshall-Smith Syndrome (MSS), characterized by accelerated skeletal maturation (often starting in utero), respiratory difficulties, mental retardation, and conspicuous physical characteristics including a long, slender neck, exophthalmos, beak-like nose, frontal bossing, and micrognathia. Our patient was a full-term infant but was noted to be small for gestational age (birth weight 2350 gm). In the newborn period the patient was noted to have several dysmorphic features consistent with MSS. Chromosomal studies were normal. No metabolic disorders were found. A skeletal survey conducted within 2 weeks of birth revealed a bone age of 4 years of age. The results of the genetic analysis and skeletal survey, along with the dysmorphic features, confirmed the diagnosis. In the neonatal period the child developed severe respiratory distress consistent with MSS requiring intubation and a subsequent tracheostomy. The patient remained tracheostomy-dependent and was home on positive pressure ventilation. As he grew older, the child’s profound mental retardation became evident. In view of the natural history of this disease, the patient had DNR orders in place signed by his family. The patient presented to our ED with a traumatically ruptured globe. After a brief period of conservative management, and in consultation with the family, the decision was made to proceed with surgical repair of the eye. The DNR orders were temporarily revoked.

The patient was brought to the OR on humidified oxygen via the tracheostomy. Standard ASA monitors were applied, a 20-gauge IV was placed and connected to a balanced salt solution containing dextrose to prevent hypoglycemia. A forced hot-air heating blanket was used to prevent hypothermia in this frail patient. Before induction, the uncuffed 5.0 Shiley tracheostomy tube was changed to a cuffed tube to allow controlled ventilation. Bronchoscopy ensured appropriate placement of the tracheostomy tube above the carina. The child was placed on pressure-mode ventilation: PAP 19 cm of water, PEEP of 5 cm of water, RR of 16 breaths/ min, FIO2 0.5. Sevoflurane 2 -3% was administered. Because the patient had a chronic well-compensated respiratory acidosis with pCO2s 50 and an HCO3 of 35, we ventilated the patient to maintain his normal pCO2 to prevent an acid-base imbalance.

Discussion: To our knowledge, this is one of the oldest patients reported with MSS coming for surgery. The incidence of MMS is not known but it is considered a rare condition. The anesthetic concerns include meticulous care to prevent respiratory decompensation and maintenance of normal homeostasis (temperature, blood sugar, padding of pressure points) in these very ill patients.