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A Case of Conradi-Hünemann Syndrome

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Conradi-Hünemann Syndrome

Conradi-Hünemann Syndrome is a rare X-linked autosomal genetic disorder that is classified as a chondrodysplasia punctate, a group of disorders characterized by the abnormal development of hardened calcium found on the heads of bones and cartilage throughout the body, where the body is still continuing to grow and develop.

Patients, primarily female, diagnosed with Conradi-Hünemann Syndrome typically present several skeletal abnormalities such as severe scoliosis, shortened limbs, malformed hips, webbing between the hands, clubbed feet, malformed hips, and deformities in the spinal column. Additionally patients may develop prominent foreheads, deformed ears, flattened cheekbones, and upturned nostrils. Along with the musculoskeletal defects, patients affected by this disease may have microphthalmia, glaucoma, and cataracts, or haziness in the lens, which often causes blurriness and may affect the clarity of their vision. In certain cases, the Conradi-Hünemann syndrome may interfere with Anesthesia due to the possibility of a shortened neck and stenosis of the trachea and the larynx.

These patients are also subject to cardiac failure, such as patent ductus arteriosus, atrial septal defects, ventricular septal defects, and pulmonary artery stenosis. Patients may exhibit renal impairment, and ventilatory failure arising from several thoracic abnormalities. When intubating the patient, it is essential to protect the patient's skin with creams and padding and monitor thermoregulation, as these patients are prone to losing heat quickly.

Case Description

A 9 year-old girl presented for Cotrel cast change for thoracolumbar scoliosis who had prior anesthetic management for similar procedures. We did an inhalational induction with oxygen and sevoflurane. After obtaining an intravenous line, anesthesia was maintained with air/oxygen mixture with sevoflurane after applying an LMA. Her head was supported with the airway maintained for the duration of the case. The cast was changed by the orthopedic surgeon while the patient was elevated on a specialized operating room table. Her anesthetic and postoperative recovery was uneventful.

Anesthetic Management

- Short neck and micrognathia in these patients may cause difficulty with direct laryngoscopy and tracheal intubation
- Laryngeal and tracheal calcifications with associated tracheal stenosis may require a smaller endotracheal tube diameter for intubation
- Upper airway obstruction can result from laryngomalacia. Patient's with this associated pathology may require positive pressure ventilation including positive end-expiratory pressure or continuous positive airway pressure to stent the airway open
- Odontoid hypoplasia can result in atlantoaxial instability and an unstable cervical spine. Manual in-line immobilization of the neck in suspected patients is required for intubation
- Ichthyosis, a skin disorder characterized by thickened, scaly skin, requires careful consideration of patient positioning to prevent unwarranted damage at pressure points. Topical agents for skin protection may prevent adhesives for vital monitors or securing IV catheters to stick onto skin. Severe Ichthyosis may impair thermoregulation.

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