Anesthesia in an infant with Lenz Majewski Hyperostotic Dwarfism. About a case.

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BACKGROUND Hereby we present the anesthetic approach in the case of a baby with several malformations in face, airway and lungs (CT scan diagnosed), included in a syndrome confirmed after surgery: Lenz-Majewsky Hyperostotic Dwarfism (LMHD).

CASE REPORT First-cousin parents. 40 days after birth the baby requires extramucosal pyloromyotomy. Upon arrival at the operating room, the first thing that draws our attention is the low weight (2.1 kg); also an anomalous phenotype with exophthalmos and ocular hypertelorism, prominent and sharp upper lip, ojival palate, large and low ears, and retrognathia. Moreover, the baby had generalized skin folds and syndactyly of 3rd- 4th fingers and 2nd- 3rd toes. The thoracic CT scan evidenced pulmonary hypoplasia and tracheal bronchus corresponding to a right superior lobar. As there was the possibility of being a difficult airway, we prepared some useful tools. Initially, we performed anesthetic induction with sevoflurane and orotracheal intubation with ETT nº 2.5; ventilation with volatile anaesthetic (sevoflurane and O₂/air 100%), maintenance with volume-controlled ventilation (tidal volume (TV) 4- 6 ml/kg) and intraoperative analgesia with intravenous fentanyl (1 mcg/kg). No neuromuscular relaxant was used. The baby was intubated after two attempts with straight shovel nº 0 (Cormack-Lehane II) and then there was a drop in the end tidal values of CO₂ (etCO₂), ranging from 24-26 mmHg in manual ventilation to 0 in controlled ventilation. After checking the ventilator, airway and blood pressure, the hypocapnia was attributed to the pulmonary malformations. The absence of etCO₂ was corrected by adjustment of TV (3 ml) and respiratory rate (from 35 to 30 breaths per minute). During surgery, SaO₂ ranged between 94-99%; we decided not to use PEEP but we performed some slight manual recruitment maneuvers. Awakening developed without incidences and the baby was transferred to pediatric ICU safely. Later, pediatricians requested a genetic study that confirmed a *de novo* heterozygous mutation of the phosphatidylserine synthase 1st gene¹ of LMHD, a very rare syndrome (prevalence < 1/1,000,000) with autosomal dominant pattern.

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KEY WORDS Dwarfism; macrocephaly; cutis laxa; syndactyly; Lenz-Majewski syndrome.

MAIN CHARACTERISTICS OF THE SYNDROME

- ☐ Craneofacial dysmorphic evolution. Large head with prominent veins in scalp, macroglosia, prognathism, midface hypoplasia, upper airway obstruction;
- ☐ Reduced trunk and limbs;
- ☐ Large fontanels, widely separated sutures;
- ☐ Large and floppy ears, choanal atresia/stenosis;
- ☐ Nasolacrimal duct obstruction;
- ☐ Cryptorchidism, inguinal hernia;
- ☐ Severe mental retardation, failure to thrive;
- ☐ Craniometaphyseal and craniodiaphyseal dysplasias.

SKELETAL ALTERATIONS

- Progressive sclerosis of the skull, facial bones and vertebrae;
- Broad clavicles and ribs;
- Short/absent middle phalanges;
- Diaphyseal undermodeling and midshaft cortical thickening;
- Metaphyseal and epiphyseal hypostosis;
- Retarded skeletal maturation;
- Defective tooth enamel.

ANAESTHETIC CONSIDERATIONS

1. Carefully evaluate airway¹ and plan for a potential difficult management²: induction with volatile anaesthetic technique, maintenance of spontaneous breathing.
2. Regional anesthetic techniques should be complemented by controlled sedation or general anesthesia.
3. Consider previous lung function test, if available.
4. Special attention in positioning the patient because of musculo-skeletal disorders diagnosed/suspected².

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