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Anesthesia in an infant with an unknown type of dwarfism. About a case

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Background

We present the anesthetic approach in the case of a baby with several malformations in face, airway and lungs (CT diagnosed), included in a very rare syndrome confirmed after surgery: Lenz-Majewski Hyperostotic Dwarfism (prevalence < 1/1,000,000).

Keywords: Dwarfism, Macrocephaly, Cutis Laxa, Syndactyly, Lenz-Majewski Syndrome

Case report

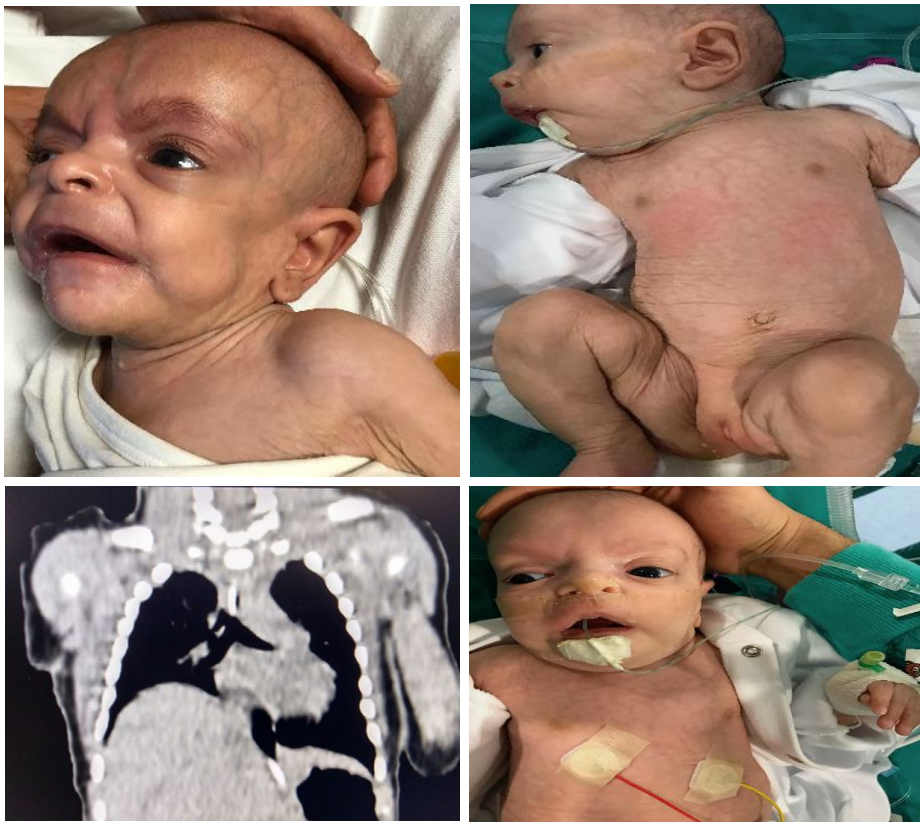
First-cousin parents. 40 days after birth the infant requires extramucosal pyloromyotomy. Upon arrival at the operating room, some features draw our attention: low weight (2,100 g), exophthalmos and ocular hypertelorism, prominent and sharpy upper lip, ojival palate, large and low ears, and retrognatia. Moreover, the baby has generalized skin folds, thin and redundant skin and syndactyly of 3rd- 4th fingers and 2nd-3rd toes. The thoracic CT evidences pulmonar hypoplasia and tracheal bronchus corresponding to a right superior lobar. The anesthetic approach was addressed as a case of difficult airway. We performed anesthetic induction with sevoflurane and orotracheal intubation with straight shovel nº 0 (Cormack II) using ETT nº 2.5, without incidences. Maintenance was made with sevoflurane, high O2 rate and fentanyl, without neuromuscular relaxants. Ventilation was based on volume-controlled mode (respiratory rate from 35 to 30 breaths/minute and tidal volume 4- 6 ml/kg depending on CO2 levels). Just after intubation there was a drop in the end tidal values of CO2, ranging from 24- 26 mmHg in manual ventilation to 0 in controlled ventilation probably due to pulmonary malformations. 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 LMHD, caused by a de novo heterozygous mutation of the phosphatidylserine synthase 1 gene with autosomal dominant pattern[1], not usually diagnosed just after birth so therapeutic treatments can be necessary before diagnosis[2].

Discussion

Clinicians should always be prepared to deal with non-diagnosed syndromic patients, observing them and getting ready to cope with difficulties that could be found depending on suspicion. For anesthesiologists, airway, cranial and hemodynamic features are very relevant[1]. Lenz-Majewski Hyperostotic Dwarfism is not usually diagnosed just after birth so patients sometimes need therapeutic treatments before diagnosis[2]. The main characteristic of the phenotype is the craniofacial dysmorphic evolution (large head with prominent veins in scalp, macroglossia, prognathism, midface hypoplasia, large fontanels, widely separated sutures, large & floppy ears, choanal atresia/stenosis, nasolacrimal duct
obstruction). Also, upper airway obstruction, reduced trunk & limbs, cryptorchidism, inguinal hernia. They may have severe mental retardation with failure to thrive; craniometaphyseal/craniodiaphyseal dysplasias and many other skeletal alterations.

Learning points

1. Carefully evaluate airway and plan for a potential difficult management[2].
2. Inhalation induction could be the safest approach in these cases.
3. Regional anesthetic techniques should be complemented by controlled sedation or general anesthesia.
4. We should always consider the previous studies done to any patient, in this case CT and lung function test were the most relevant ones.
5. Assessment of malformations is crucial for the best anesthetic management.
6. Special attention in positioning the patient specially if muscle-skeletal disorders diagnosed or suspected[2].



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