



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 sharp upper lip, ovoid palate, large and low ears, and retrognathia. 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 pulmonary 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 n° 0 (Cormack II) using ETT n° 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 pattern¹, not usually diagnosed just after birth so therapeutic treatments can be necessary before diagnosis².

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 relevant¹. Lenz-Majewski Hyperostotic Dwarfism is not usually diagnosed just after birth so patients sometimes need therapeutic treatments before diagnosis². 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 management².
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 suspected².



References

1. Wattanasirichaigoon, D., Visudtibhan, A., Jaovisidha, S., Laothamatas, J., Chunharas, A. Expanding the phenotypic spectrum of Lenz-Majewski syndrome: facial palsy, cleft palate and hydrocephalus. *Clin. Dysmorph.* 13: 137-142, 2004.
2. Victor C Baum MD, Jennifer E. O'Flaherty MD MPH; *Anesthesia for Genetic, Metabolic and Dysmorphic Syndrome of Childhood*. 2nd Ed. Philadelphia: Lippincott Williams & Wilkins; 2006.
3. Sousa SB, Jenkins D, Chanudet E, Tasseva G, Ishida M, Anderson G, Docker J, Ryten M, Sa J, Saraiva JM, Barnicoat A, Scott R, Calder A, Wattanasirichaigoon D, Chrzanowska K, Simandlova M, Van Maldergem L, Stanier P, Beales PL, Vance JE, Moore GE. Gain-of-function mutations in the phosphatidylserine synthase 1 (PTDSS1) gene cause Lenz-Majewski syndrome. *Nat Genet.* 2014 Jan;46(1):70-6. doi: 10.1038/ng.2829. Epub 2013 Nov 17
4. Wattanasirichaigoon D, Visudtibhan A, Jaovisidha S, Laothamatas J, Chunharas A. Expanding the phenotypic spectrum of Lenz-Majewski syndrome: facial palsy, cleft palate and hydrocephalus. *Clin Dysmorphol.* 2004 Jul;13(3):137-42
5. Gorlin RJ, Whitley CB. Lenz-Majewski-syndrome. *Radiology.* 1983 Oct;149(1):129-31
6. Robinow M, Johanson AJ, Smith TH. The Lenz-Majewski hyperostotic dwarfism. A syndrome of multiple congenital anomalies, mental retardation, and progressive skeletal sclerosis. *J Pediatr.* 1977 Sep;91(3):417-21
7. Lenz WD, Majewski F. A generalized disorders of the connective tissues with progeria, choanal atresia, symphalangism, hypoplasia of dentine and craniodiaphyseal hypostosis. *Birth Defects Orig Artic Ser.* 1974;10(12):133-6
8. Victor C Baum MD, Jennifer E. O'Flaherty MD MPH. *Anesthesia for Genetic, Metabolic, and Dysmorphic Syndromes of Childhood*. 2nd edition, Philadelphia: Lippincott Williams & Wilkins; 2006
9. Bruno Bissonnette, Igor Luginbuehl, Bruno Marciniak, Bernard Dalens. *Syndromes – Rapid recognition and perioperative implications*. McGraw-Hill, New York; 2007.
10. A Japanese patient with a mild Lenz–Majewski syndrome. Sumito Dateki Æ Tatsuro Kondoh Æ Gen Nishimura Æ Katsuaki Motomura Æ Koh-ichiro Yoshiura Æ Akira Kinoshita Æ Hideo Kuniba Æ Yoshiyuki Koga Æ Hiroyuki Moriuchi. Received: 10 April 2007 / Accepted: 17 May 2007 / Published online: 26 June 2007. The Japan Society of Human Genetics and Springer 2007 *J Hum Genet* (2007) 52:686–689 DOI 10.1007/s10038-007-0165-y