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Case report

Cephaloskeletal dysplasia (Taybi-Linder syndrome): Case report and anesthetic considerations☆

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ABSTRACT

Introduction: Microcephalic osteodysplastic primordial dwarfism (or Taybi-Linder syndrome) is a rare disease characterized by bone and central nervous system malformations, in addition to intrauterine retardation.
Case presentation: 20-year-old patient operated on for adhesiolysis and enteropexy due to bowel obstruction from post surgical adhesions.
Conclusion: The anesthetic considerations in these patients include the potential airway impairment secondary to facial malformations and neurological complications, primarily seizures.

Displasia cefaloesquelética (Síndrome de Taybi-Linder): Presentación de un caso y consideraciones anestésicas

Palabras clave:
Enanismo
Anestesia, niño
Manejo de la vía aérea
Osteocondrodisplasia

RESUMEN

Introducción: El enanismo microcefálico osteodisplásico primario (o síndrome de Taybi-Linder) es una infrecuente enfermedad caracterizada por malformaciones óseas, del sistema nervioso central y crecimiento intrauterino retardado.
Presentación del caso: Paciente de 20 años intervenida de adhesiolisis y pexia intestinal por un cuadro de obstrucción intestinal por bridas postquirúrgicas.

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Conclusión: Como consideraciones anestésicas de estos pacientes destacamos las posibles alteraciones de la vía aérea secundarias a las malformaciones faciales y las complicaciones neurológicas, principalmente crisis convulsivas.

Introduction

The Taybi-Linder syndrome, also known as microcephalic osteodysplastic primordial dwarfism (MOPD) types I and III, is a rare, potentially inherited autosomal recessive condition. It is characterized by intrauterine growth retardation (IGR), bone dysplasia (including the facial skeleton) and central nervous system malformations including brain malformations, refractory epilepsy, sensitive deficits, cognitive deficits and neuroendocrine disorders. Notwithstanding the potential complications that the anesthesiologist may face, primarily related to airway abnormalities and the frequent association with neurological pathology, particularly epilepsy and neuroendocrine disorders, there are very few publications in the specialized bibliography.

Majewski et al.[1,2] classified patients with microcephalic primordial dwarfism into two categories: Seckel syndrome and osteodysplastic primordial dwarfism (ODPD) that is further subdivided into three groups: I, II and III, primarily based on the skeletal characteristics. Seckel syndrome was defined as severe IGR and postnatal dwarfism with severe microcephaly characterized by "bird-like face", micrognathia, pointed nose, and receding forehead, associated with cognitive impairment and other abnormalities but with no skeletal malformations, except for occasional dislocation of the head of radius. Patients with ODPD present the same craniofacial characteristics but associated with skeletal abnormalities. The current medical bibliography equates the definition of Taybi-Linder syndrome with ODPD types I and III.

The situations when the anesthesiologist may be faced with this pathology range from complementary testing (imaging diagnosis), mainly in pediatric patients, to perioperative management of anesthesia, primarily orthopedic surgery as a palliative approach to bone abnormalities.

Clinical case

Patient information

This case refers to a 20-year-old patient (10 kg of body weight), undergoing exploratory laparotomy due to bowel obstruction from adhesions. The patient was diagnosed with Taybi-Linder syndrome and total colonic Hirschsprung requiring total colectomy and ilio-anal anastomosis, in addition to a number of subsequent re-interventions due to obstructions secondary to intestinal adhesions. There were no difficulties with the endotracheal intubation of the patient during the previous procedures.

Clinical findings and interventions

A detailed history of the patient was recorded during the preanesthesia consultation before surgery. Due to poor patient cooperation, there was some degree of difficulty to evaluate the airway, the absence of epileptic seizures in the last 5 years (without basal treatment), or any hydro electrolytic imbalances. No difficult airway predictors or difficult manual ventilation were identified during the examination of the airway; however, the patient's facial anatomy was suggestive of difficult intubation. Based on past surgical experiences with no difficulty to access the airway, no special measures were taken in this regard.

The patient arrived at the OR with adequate anxiolysis (2 mg of intranasal midazolam administered 30 min earlier) to proceed with inhaled induction (8% sevoflurane with fresh gas flow of 6 L/min and tidal volume for spontaneous breathing) while standard monitoring procedures are followed (electrocardiography, non-invasive blood pressure control, and pulse oximetry), in addition to pediatric bilateral bispectral index (BIS), including spectral density matrix (SDM) monitoring. A peripheral 22 G venous line is inserted in the right upper limb for the administration of 30 mg of Propofol, 30 μg of fentanyl, and 10 mg of rocuronium. After completing the anesthetic induction, the orotracheal intubation is performed with no preliminary manual ventilation (5.0 mm external diameter tube with pneumoplugging) for full visualization of the glottis, Cormack–Lehane Grade I.

Anesthesia is maintained with sevoflurane 0.8 CAM (BIS target 40–60) in semi-closed loop (fresh gas flow 1 L/min, 40% inspired oxygen fraction in medicinal air), and fentanyl dosing as needed for adequate analgesia. The procedure begins with a supra and infra umbilical laparotomy approach to proceed with the adhesiolysis and enteropexy. Support vasoactive treatment and blood products transfusion were not required during the intervention. No remarkable changes were observed in the BIS monitor EEG wave, with no evidence of SDM waves or registry compatible with epileptic seizures.

The postoperative analgesia was administered using a morphine chlorine pump for 48 h and NSAIDs.

Follow-up

When the surgical procedure was over, the patient was educted from anesthesia and extubated in the OR uneventfully. The clinical evolution of the patient was