Rare disease

Administration of general anaesthesia to a paediatric patient with osteopetrosis

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Summary

Osteopetrosis is a rare clinical syndrome characterised by the failure of bone resorption and remodelling, which causes multiple anatomical and physiological impairments. Pathological fractures can occur, in addition to, haemathological and metabolic impairments. Our patient was a 9-year-old girl diagnosed with osteopetrosis in the neonatal period. She had severe anaemia, thrombocytopenia, hypocalcaemia, as well as growth and development delays. In this case report, the administration of general anaesthesia to the patient for a biopsy of the scalp and skull and a partial maxillectomy is presented.

BACKGROUND

Osteopetrosis (also known as osteosclerosis or marble bone disease) is a rare disease, characterised by the failure of bone resorption and remodelling which leads to multiple anatomical and physiological impairments. Different forms of osteopetrosis occur based on inheritance patterns.^{1 2} These are type I and II autosomal dominant osteopetrosis as well as benign and malignant autosomal recessive osteopetrosis. Infantile malignant osteopetrosis is autosomal recessive inherited, usually affects children less than 1 year old, and has a poor prognosis. Pathological bone fractures can occur in addition to respiratory, neurological and haematological impairments.^{3 4}

CASE PRESENTATION

A 9-year-old girl patient with osteopetrosis was admitted to the paediatric clinic due to severe anaemia. The biopsy and maxillectomy were planned by the otorhinolaryngologists and the neurosurgeons in response to necrotic areas on the patient's scalp and palate. On preoperative examination, the patient weighed 14 kg (<3 percentile) and her height was

92 cm (<3 percentile). She had growth and development delays, abdominal distension (due to hepatosplenomegaly), necrotic wounds in her oral cavity and cranium, exophthalmia, hypertelorism, bilateral optic atrophy, hearing impairment and micrognathia (figures 1–2). Chest auscultation revealed bilateral rales. Her ability to open her mouth and to extend her head were limited. The patient was prescribed methyl prednisolone, metronidazole, ampicillin and ceftriaxone. Her echocardiogram was normal. The head CT scan showed pneumo-hydrocephalus, and displaced segmental fractures on her nasal bone, ethmoid bone, right zygomatic arch, lateral, medial and inferior walls of the orbit, and hard palate. The zygomatic suture was separated. Her laboratory findings were as follows: white blood cells: 6 060/mm³ (4600–10 200); haemoglobin (Hb): 6 g/dl (12.2–18.1); Hematocrit (Htc): 17.6% (37.7–53.7); platelets (Plt): 36 000 (142 000–424 000); Na: 132 mEq/l (135–145); K: 3.3 mEq/l (3.5–5.5); Ca: 6.6 mEq/l (8.5–10.8); blood sugar: 103 mg/dl (75–115); urea/creatinine: 15/0.4 mg/dl (10–50/0.6–1.2) and aspartate aminotransferase/alanine transaminase 9/4 U/l (5–40/5–40). The patient was administered 7 U platelet suspension (5×10¹⁰ platelet count per unit), 2 U erythrocyte suspension, 1 U freshly frozen plasma and calcium before the operation. On a second control blood count, her results were as follows: Hb: 10.4 g/dl; Htc: 30.4%; Plt: 99 000; and Ca: 8.4 mEq/l. A consent form for invasive airway manoeuvre was signed by the patient's relatives because a difficult intubation was anticipated. One unit of platelet suspension was prepared to administer during the operation. The patient's continuous three-lead ECG, non-invasive blood pressure and peripheral oxygen saturation were monitored. Her heart rate was 130 beats/min with an arterial blood pressure of 89/60 mmHg and an SpO₂ of 94% on room air. Endotracheal tubes, blades (0, 1 and 2 number, both Miller and Macintosh type) and masks in different sizes were prepared. Anaesthesia mask induction with sevoflurane, without using muscle relaxants, was planned because a difficult intubation was suspected. Anaesthesia was maintained with 2.5% sevoflurane in 50% O₂–50% air. During attempt intubation they were

Figure 1 The patient's general appearance.

Figure 2 The side view of the patient.

determined that head extension was limited, chin was small and the vocal cords were not seen (Cormack-Lehane Grade 3).⁵ After two unsuccessful attempts with a Macintosh laryngoscopy blade, we changed to a Miller blade, size 1. We were able to visualise the patient's vocal cords (Cormack-Lehane Grade 2) and an endotracheal tube (inner diameter, 4 mm) was passed with bilateral breath sounds and end tidal carbon dioxide wave. The operation lasted approximately 30 min. Postoperation, the patient was extubated and taken to the clinic fully awake.

DISCUSSION

Although pathological fractures are the most common clinical manifestation of osteopetrosis, respiratory, neurological, haematological and metabolic impairments can occur. Despite this excess bone formation, people with osteopetrosis tend to have bones that are more brittle than normal. Mild osteopetrosis may cause no symptoms, and present no problems. However, serious forms of osteopetrosis can result in stunted growth, deformity, increased likelihood of fractures, and also patients suffers from anaemia, recurrent infections and hepatosplenomegaly due to bone expansion leading to bone marrow narrowing and extramedullary haematopoiesis. It can also result in blindness, facial paralysis and deafness, due to the increased pressure put on the nerves by the extra bone.^{6 7} The operation on the patient's oral cavity, scalp and bones was planned. The scalp has good blood circulation and, therefore, is prone to bleeding.⁸ Moreover, surgery on the oral cavity and tissue trauma due to endotracheal intubation can cause continuous bleeding from the operation site, resulting in tracheal aspiration.^{9 10} Because major bleeding was expected during the surgery, the patient's haematological parameters needed to be improved by transfusion. The patient received 2 U erythrocyte and 7 U platelet suspensions to normalise her

haematological parameters prior to surgery. Hb increased from 6 to 10.4 g/dl and platelet count increased from 36 000 to 99 000.

Ventilation of the lung is another area of concern for paediatric patients with osteopetrosis. Endotracheal intubation may be difficult due to abnormalities of the head and neck, including high and narrow palate, mandibular widening and hypoplasia, temporomandibular joint restriction and fractures of the cervical vertebrae.^{11–13} Moreover, distention of the abdomen due to hepatosplenomegaly can cause eventration of the diaphragm, resulting in difficulty with ventilation.¹⁴ Our patient had the aforementioned head and neck anomalies, ventilation was without problem, but the intubation was successful on the third attempt. As noted in the American Society of Anesthesiologists difficult airway algorithm, the anaesthesiologist should have a preformulated strategy for intubation of the difficult airway. First if possible, awake intubation should be preferred. If this is not possible and the initial attempt at endotracheal intubation is not successful, then calling for assistance, change of equipment and operator are all recommended. If possible return to spontaneous ventilation should be attempted. Alternatively placement of a laryngeal mask airway (LMA) should be considered. If possible the surgery can be completed with an LMA in place or the LMA can be used to facilitate endotracheal intubation. However, if these measures are all unsuccessful a surgical airway may be necessary. As the procedures planned for our patient precluded the use of an LMA, endotracheal intubation was necessary.¹⁵ With these kinds of patients, it is important to be prepared for any condition; preparations should be made for performing an emergency invasive airway manoeuvre. Furthermore, instead of an invasive approach, such as endotracheal intubation, the use of LMA is recommended.¹⁶ However, since mask ventilation was considered difficult for our patient and the procedure was in oral cavity, endotracheal intubation

was preferred to prevent tracheal aspiration. In addition, if a difficult intubation is suspected or ventilation is problematic, muscle relaxants should be used with caution until the airway is secured.

Our patient had pneumocephalus, so nitrous oxide was not used during anaesthesia; because nitrous oxide will diffuse into air filled spaces and in this case increasing the size of the pneumocephalus.¹⁷

Infantile malignant osteopetrosis can cause imbalanced calcium homeostasis, such that, despite the presence of sufficient amounts of calcium and phosphorus in the body, bone mineralisation is impaired.^{18 19} Our patient also had hypocalcaemia and received calcium prior to surgery.

Patients with osteopetrosis suffer from difficult ventilation and intubation due to bone defects in addition to severe haematological and metabolic impairments. Thus, the management of these patients must include a thorough evaluation of the affected systems prior to surgery, and anaesthesia application must be planned accordingly.

Learning points

- ▶ Mask ventilation and endotracheal intubation may be difficult. Endotracheal intubation should be anticipated as difficult and the necessary precautions taken.
- ▶ Abnormal electrolyte and haematological parameters should be treated in anticipation of further abnormalities associated with the anticipated surgery.
- ▶ Osteopetrosis is associated with pneumocephalus, hence nitrous oxide should be avoided.

Competing interests None.

Patient consent Obtained.

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