Case Report

Pycnodysostosis and severe laryngomalacia complicating general anesthesia: A case report

ABSTRACT

Pycnodysostosis is a very rare autosomal recessive disease. This disorder presents with osteosclerosis, leading to fragile bones, short stature, craniofacial abnormalities, laryngomalacia, sleep apnea syndrome, and many other findings. Difficulty intubation is very common. In our case, we have a patient scheduled for a plastic surgery on her skull under general anesthesia (GA), but she could not lie in a flat position as this could lead to cyanosis and cardiac arrest. So, we have to intubate her in 45° back-up position. Intubation in positions different from the usual supine one probably because of severe laryngomalacia in this patient is possible but needs skilled anesthesiologists and trained personnel.

Key words: Bone fractures, cardiac arrest, endotracheal intubation, laryngomalacia, patient positioning, pycnodysostosis

Introduction

Pycnodysostosis (Toulouse-Lautrec syndrome or Maroteaux–Lamy syndrome) is a very rare autosomal recessive disease that presents with osteosclerosis, leading to fragile bones, increased risk of fractures after minor trauma, short stature, craniofacial abnormalities, laryngomalacia, sleep apnea syndrome, and difficulty intubation. Stridor as a cause of laryngomalacia seems to be very common (20%), and we report here a case of a patient who could not lie down at all times. She had to stay in a 45° back-up position because she risks having respiratory difficulty leading to cardiac arrest. In cases of general anesthesia (GA), she had to be intubated in this position as we show in one picture.

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Case Report

After patient consent form has been obtained from a 65-year-old woman scheduled for a surgery on her skull (a cutaneous flap). She had the diagnosis of pycnodysostosis after multiple fractures in her past all of them treated surgically under regional anesthesia. She had short stature, deformed articulation, and some signs of difficult intubation: Mallampati 3, interincisor distance equal to 2,0 cm, large tongue, and thyromental distance equal to 4 cm [Figure 1]. Her laboratory and cardiac exams were under normal limits. The main problem: she had a respiratory stridor and was very uncomfortable when in supine position. She was submitted to GA in two episodes previously. In one of them,

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Plínio O. Holanda, Luciana C. Morais, Felipe O. Marques, Danielle C. Soares, Joao M. R. Neto, Keyth W. B Silva

Anesthesiology Departament, Instituto José Frota, Fortaleza, Ceará, Brazil

Address for correspondence: Dr. Plínio O. Holanda, Rua Barão Do Rio Branco 1816, Fortaleza, Brazil.
E-mail: anesthesiaceara@gmail.com

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she had a substantial desaturation just after induction of anesthesia and also an extubation failure in supine position. A tracheostomy was undertaken as an urgent procedure. A nasal endoscopy to assess the trachea was inconclusive because the tracheostomy was still in place. In another moment, she had cardiac arrest just before induction of anesthesia and after extubation, both in supine position. Thereby, after these episodes it seemed probable that the reason was a respiratory issue and we supposed she had an important laryngomalacia. Although she had signs of difficult intubation, this was not a problem during the cardiac events and we were not worried about this. Even though, an awake intubation in 45° back-up was prepared this was probably the best position for her. One milligram of midazolam plus dexmedetomidine 1 mcg/kg/h IV was administered. During the preparation for the awake intubation, she reacted very badly to the laryngeal topical anesthesia: her stridor worsened very much and she became confused. So, we decided to move to GA in 45° back-up position [Figure 2] using fentanyl 100 mcg + etomidate 10 + 10 mg intravenous after preoxygenation with 10 L/min oxygen with a bag-valve-mask device for three minutes. Tracheal intubation was very easy despite the position. Maintenance of anesthesia was made with sevoflurane and nitrous oxide. There were no events during the procedure, and she was extubated in this position with no complaints. After 2 hours in the recovery room, she was transferred to the ward asymptomatically.

Discussion

Pycnodysostosis (Toulouse-Lautrec syndrome or Maroteaux–Lamy syndrome) is a lysosomal skeletal dysplasia that is autosomal recessive. It was individualized by Marteau and Lamy in 1962. The individual with this syndrome is phenotypically characterized by short stature, typical facial appearance (small jaw with obtuse mandibular angle and convex nasal bridge), stridor, laryngomalacia (20%), obstructive sleep apnea (>60%), and difficult intubation.[1,2] Increased bone density, acro-osteolysis, and an increased risk of fractures after minor trauma are common. Incidence is about 1.7 per 1 million births[3] and approximately 200 affected individuals have been reported in the medical literature.[4] Craniofacial defects include frontal and parietal bossing, beaked nose, prominent eyes with blue sclera, hypoplastic maxilla and mandible, depressed nasal bridge, open fontanels and sutures, thick calvaria, hypoplastic paranasal sinuses, high-arched grooved palate, and elongated soft palate resulting in snoring and mouth breathing. Clinical heterogeneity with its rarity challenges the prognosis in these patients. Some cases of airway obstruction associated with pycnodysostosis reported in the literature are situated at various levels of the airways[4], and stridor is reported[5] probably related to laryngomalacia that can lead to complete upper airway obstruction and cardiac arrest when in supine position. Attempting awake intubation with lidocaine in patients attained of laryngomalacia seems to get worse the already present obstruction as described by some authors but questionable by others,[6,7] so the best option for us was to intubate her in a 45° back position under GA. There is a case report from Japan where a patient was intubated in this position.[8] In our case, this was made uneventfully. Two days after the operation, the patient was transferred home without any discomfort. Now, she and her family are aware of her problem and may help other doctors in case GA is necessary for this fragile patient.

Conclusion

Pycnodysostosis is a challenging syndrome that can lead to bone fractures easily and difficult intubation. Our patient had another problem: She could not breathe in supine position

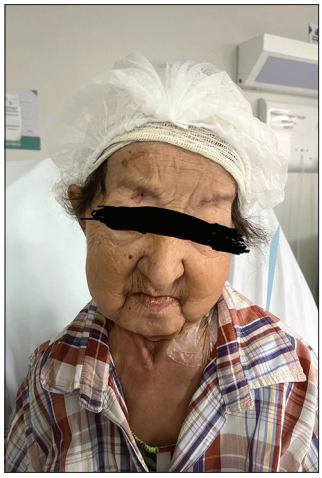

Figure 1: Patient with pycnodysostosis

Figure 2: Intubation in 45° back-up

maybe because of an important laryngomalacia, so the only option was to intubate her in a 45° position. We did not find another similar case in the literature. In case she needs another intubation, this will be probably necessary.

Written informed consent was obtained from the patient's guardian.

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Conflicts of interest
There are no conflicts of interest.

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