

Pycnodysostosis: Anaesthetic Challenges

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Received: August 2019

Accepted: August 2019

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ABSTRACT

A rare autosomal recessive disorder Pycnodysostosis is to mutation of CTSK gene. This group of patients have fragile bone, craniofacial abnormalities with difficult airway. We are describing the anaesthetic management of a 12 yr male with pycnodysostosis for lower limb fracture fixation in combined spinal epidural anaesthesia.

Keywords: Pycnodysostosis, Osteopetrosis, Capthesin gene.

INTRODUCTION

Pycnodysostosis (also known as Toulouse-Lautrec syndrome) is a rare inborn error with defective activity of osteoclast.^[1,2] The disease manifests as short stature, craniofacial deformity, generalized osteosclerosis, dysplastic bones including hypoplasia of the distal phalanges, clavicles, and various craniofacial bones.^[3,4] Patients of these disease are generally referred for surgical intervention due to upper airway obstruction, pathological fracture or any dental complications.

Prevention of the above complications and growth hormone therapy for stunted growth are the management strategies.

CASE REPORT

A 12 year male child is referred for surgical correction of closed traumatic fracture of shaft of femur of right side after fall from stairs. The patient had a history of fracture of shaft of femur of opposite side and surgical intervention was done which was uneventful. He has no history of any other illnesses or any allergy to any alleged drugs. On examination patient had large protruding tongue with high arched palate. Patient had Modified Mallampati class III with no dental abnormalities. He had short stature with height 145cm.

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Preoperative Blood investigations:

HB:15.1g/dl, WBC-7300/mm³, Total Platelet count: 236x10³/mm³, serum sodium-137mEq/L, potassium-4.4 mEq/L, Serum creatinine-0.4mg/dl, blood urea-44 mg/dl

PT-13.8 sec, APPT-25.6 sec, INR-1.28

After securing IV access with 18 G cannula preloading with 300 ml of Ringer Lactate was done. Standardized Monitors like electrocardiograph, Heart rate SPO₂ and Blood pressure were attached. All vitals were within normal limits before the procedure. Antibiotics Ceftriaxone 1gm was given. Under all aseptic percussion combined spinal epidural was performed at the level of L3-L4 intervertebral space with 2.4 ml of 0.5% bupivacaine heavy and 10 microgram fentanyl was injected. Epidural catheter was threaded. Appropriate Position was made after achieving a spinal level of T10. All pressure points were padded. Total duration of surgery was 130 minutes. Intraoperative period was uneventful

Post-operative analgesia was maintained by epidural bolus doses of Bupivacaine. Epidural catheter was removed after 48 hours after surgery.

DISCUSSION

Pycnodysostosis also called Toulouse-Lautrec syndrome was first defined by Mareteux and Lamy in 1962.^[5] It's a rare autosomal recessive disorder with incidence of 1 in 1.7 million population.^[6] The disease is attributed to chromosome 1q21 with defect in Capthesin K (CTSK gene), lysosomal cysteine proteases.^[7] CTSK gene which is responsible for production of enzyme capthesin is mutated in this disease. Capthesin protein helps in breakdown of proteins like bone matrix, collagen

etc. There is accumulation of protein in bone matrix due to defective gene CTSK.

The distinctive facial features of this disease are frontal bossing, separate cranial suture, beaked nose, high arched palate, bluish sclerae, and hypoplasia of the maxilla and mandible.^[3] Affected individuals may suffer from delayed eruption of tooth and brittle like bones which are more prone to fracture with minimal trauma or no trauma.^[8] Other associations are Short stature (generally <150 cm) mostly limbs, Clavicular dysplasia, acroosteolysis (in which fingers are stubby and reduced in size with absence of nails), scoliosis and chest deformity. These patients are more prone for obstructive sleep apnea. Differential diagnosis of Pycnodysostosis are following:

1. **Osteopetrosis:** Some features like delayed closure of cranial sutures and phalangeal, or clavicle hypoplasia are not noticed.^[9]
2. **Cleidocranial dysplasia:** Autosomal dominant inheritance; open fontanels and cranial sutures in advanced age with no clavicle or pharyngeal hypoplasia.^[10]
3. **Osteogenesis imperfecta (OI):** Severe multiple bone fractures as compared to pycnodysostosis. Most of the cases are associated with blue sclera and choanal atresia.^[11]
4. **Hajdu-Cheney syndrome (HCS):** mutations in the NOTCH2 gene with autosomal dominant inheritance. osteolysis and acroosteolysis are the main features of this disease.



Figure 1: Showing patient with Pycnodysostosis large protruding tongue.

Prenatal diagnosis is performed by demonstration of mutations in Capthesin K gene in chorionic villi biopsy or amniotic cells. The diseases can be suspected when characteristic features present with evaluation of family history and radiological examination.

There are many case reports describing the clinical features of the disorder, but there is paucity of

materials emphasizing the anaesthetic management in these patients.



Figure 2: Showing fracture of limb.

The main challenge for anesthesiologists is anticipated difficult airway due to distinctive facial features which makes regional anaesthesia as a preferred choice over general anesthesia when indicated. Fragile bones prone to fracture during positioning after anaesthesia. Subclavian venous cannulation becomes difficult due to clavicular dysplasia. The association of obstructive sleep with this group of patients is a risk factor for airway obstruction in post operative period. So, care should be taken to avoid intravenous opioids and patient should be monitored vigilantly in postoperative period.

Detailed airway assessment, preparation for management of difficult airways and vigilant during positioning as well as post-operative period are some key to manage this group of patients.

CONCLUSION

We are concluding that detailed airway assessment, preparation for management of difficult airways and vigilant during positioning as well as post-operative period are some key to manage this group of patients.

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How to cite this article: Barma MD, Das AK. Pycnodysostosis: Anaesthetic Challenges. *Ann. Int. Med. Den. Res.* 2019; 5(6):AN31-AN33.

Source of Support: Nil, **Conflict of Interest:** None declared