Ain-Shams Journal of Anesthesiology

CASE REPORT

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Contextual anaesthesia for a rare case of pycnodysostosis with myriad presentation: a case report

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Abstract

Background: Pycnodysostosis is a rare inherited syndrome characterized by classical craniofacial defects and diffuse osteosclerosis disorder of bones with tendency for fractures after minor trauma.

We describe the anaesthetic management of a parturient with pycnodysostosis with dual concerns of precious pregnancy and a fractured femur.

Case presentation: A 33-year-old primigravida with 31 weeks of gestation presented with subtrochanteric fracture of left femur. Clinico-radiological features of diffuse osteosclerosis suggested a diagnosis of pycnodysostosis, which was confirmed by genetic testing. We encountered challenges for sequential surgeries of lower segment caesarean section, surgical fixation of the fractured femur and surgical wound debridement in this patient in view of a difficult airway, short stature, physiological changes of pregnancy, difficulties in positioning and possibility of increased blood loss. A combination of general anaesthesia with either neuraxial anaesthesia or regional block was performed, which helped in the safe management of this syndromic patient.

Conclusions: Multidisciplinary team dynamics, communication and expertise played a crucial role in the successful care of the patient. Planning anaesthesia contextually for each surgical setting was vital. Anaesthetic gadgets like fibre-optic bronchoscope and ultrasound were a boon to anaesthesiologist at every surgical procedure to aid the anaesthetic plan.

Keywords: Contextual anaesthesia, Difficult airway, Fracture femur, Pycnodysostosis, Pregnancy

Background

Pycnodysostosis is a lysosomal skeletal dysplasia which is autosomal recessive, and the individual is phenotypically characterized by short stature, increased bone density, acro-osteolysis, mid-face hypoplasia and an increased risk of fractures after minor trauma (Turan 2014; Puri et al. 2013; Kiran et al. 2008). We present this rare case with dual concerns of pregnancy and fracture femur. To our knowledge, the anaesthetic management of pycnodysostosis for a series of surgeries in the same patient with pregnancy and femur fracture is not described in the scientific literature.

The institutional review board of our Bangalore Baptist Hospital has approved the case report, and written informed consent is taken from the patient.

Case presentation

A 33-year-old primigravida with 31 weeks of precious gestation presented to the emergency department with severe left hip pain and unable to bear weight for 1 month. Radiograph revealed a subtrochanteric fracture of the left femur. The antenatal scan showed severe oligohydramnios with an amniotic fluid index of 3.5.

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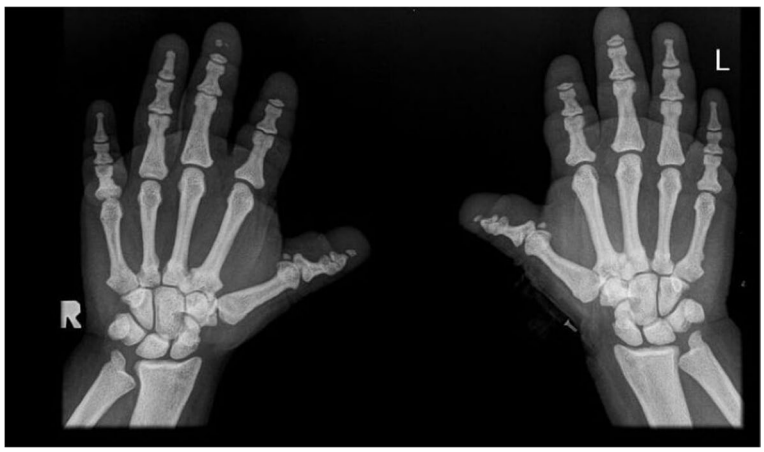

Fig. 1 Short and stubby hands with distal phalanx acro-osteolysis of thumb, index and middle fingers of both hands

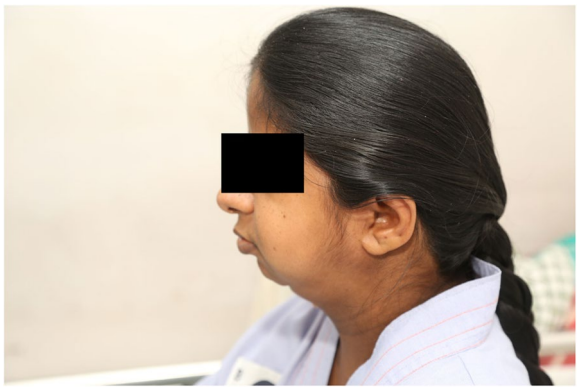

Fig. 2 Lateral profile with retrognathia and micrognathia

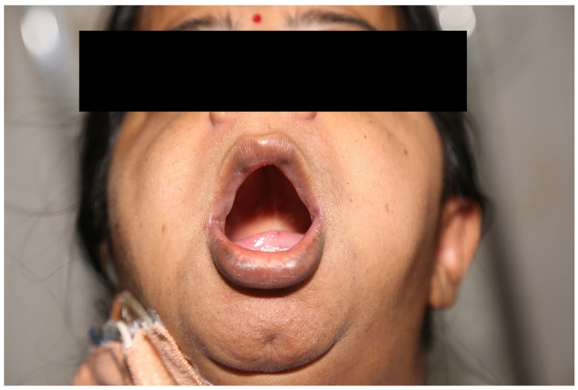

Fig. 3 Mouth opening with high-arched palate

On examination, she was short statured with a height of 137 cm and weighed 50 kg. Associated features were acromelia, short stubby hands (Fig. 1) and feet, frontal bossing and a prominent nose. Higher mental function was normal. Airway examination revealed a large protruding tongue with severe retrognathia and micrognathia (Fig. 2), thyromental distance of 3 cm, short neck, Mallampati grade 4, high-arched palate (Fig. 3), mouth opening was 2.5 cm, upper lip bite test of grade 3 and flat face. The patient's skeletal imaging showed generalized sclerosis of bones and Erlenmeyer flask deformity of the femur (Fig. 4); skull radiograph showed unfused cranial sutures, the presence of Wormian bone, and obtuse mandibular angle (Fig. 5). Radiological assessment of the spine was deferred in view of pregnancy. She was evaluated by a team of obstetrician, orthopaedician, anaesthesiologist and geneticist.

The clinico-radiological features were consistent with diffuse sclerosing bone disorder, probably pycnodysostosis. Genetic testing of the patient revealed autosomal recessive homozygous missense variation in exon 6 of the CTSK gene with a variant of c.629G > T (p. Cys210Phe) pycnodysostosis.

Precious pregnancy and high risk of bleeding deferred the decision of immediate surgical fixation on the femur. An elective lower segment caesarean section (LSCS) at 34 weeks and femur fixation as a second-stage procedure to allow time for foetal lung maturity were planned. She was treated with L-arginine, subcutaneous enoxaparin 40 mg along with skin traction and bed rest. However, during this course of treatment, she developed premature rupture of membranes and urinary tract infection with pan-resistant *Klebsiella*. An early elective LSCS was planned at 33 weeks gestational age following a course of antibiotics, steroids for foetal lung maturity and magnesium sulphate for foetal neuroprotection.

Laboratory blood investigations and transthoracic echocardiography were normal.

In view of difficulty in positioning and short stature for spinal anaesthesia (SA) and definitive difficult airway, we

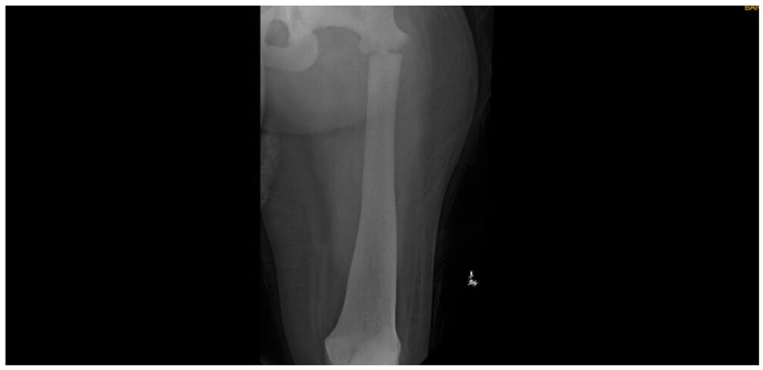

Fig. 4 Erlenmeyer flask deformity of left femur with subtrochanteric fracture

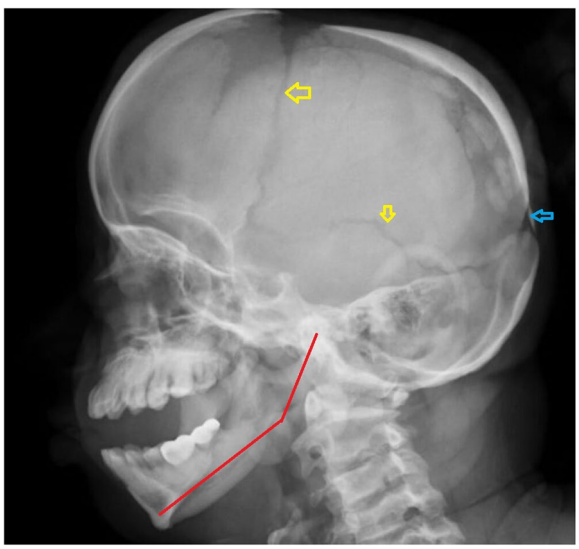

Fig. 5 Lateral radiograph of the skull showing open sutural lines (yellow lines), the presence of Wormian bone (blue line) and obtuse mandibular gonial angle (red line)

planned general anaesthesia (GA) with awake fibre-optic intubation (FOI) for LSCS. The above procedure was clearly explained to the patient.

Premedication with intravenous (IV) ranitidine 50 mg and metoclopramide 10 mg was given.

Two wide bore cannulae 18 G were secured, and bilateral superior laryngeal nerve blocks and transtracheal injection were performed with 2% plain lignocaine for awake FOI. Lignocaine 15% spray was used in each nostril and oropharynx. Awake nasal FOI with 6.5-sized tracheal tube was secured after confirming bilateral equal air entry with capnography.

Anaesthetic induction was done with propofol 1.5 $mg.kg^{-1}$ and neuromuscular block with atracurium 0.5 mg/kg. IV paracetamol 1 g was given following induction. Maintenance of anaesthesia was achieved with oxygen and air (50:50) and sevoflurane. A 1.8 kg female baby was delivered. Analgesia was provided with IV fentanyl 2 $\mu g.kg^{-1}$ and morphine 3 mg, ultrasound (USG)-guided bilateral transversus abdominis plane block, and left femoral nerve block was done with 0.25% bupivacaine with dexamethasone 8 mg with 15 ml at each site for post-operative analgesia. She was extubated, and perioperative period was uneventful.

Surgical fixation of left fracture femur was planned 2 weeks post LSCS. Concerns were prolonged surgical time, risk of blood loss and infection. Repeat blood investigations and spine radiograph were normal (Fig. 6).

GA and lumbar epidural analgesia were planned. An 18 G IV cannula and left radial arterial line were secured. The lumbar epidural catheter was placed in a sitting position. Awake oral FOI with 6.5 size cuffed tracheal tube was done with "spray-as-you-go" technique. IV induction was done with fentanyl 2 $\mu g.kg^{-1}$, propofol 2 $mg.kg^{-1}$ and atracurium 0.5 $mg.kg^{-1}$. Under USG guidance, central line was secured in the right internal jugular vein post-induction. Maintenance of anaesthesia was done with O2: air (50:50) and isoflurane. Intraoperatively analgesia was provided with 3 ml bolus doses of 2% lignocaine epidurally and IV fentanyl 0.5 mcg/kg. Mean blood pressure was kept around 60 mmHg. Blood loss was 300 ml over a duration of 5 h surgery. Postoperative analgesia was managed with 0.1% bupivacaine with fentanyl 2 $\mu g.ml^{-1}$ at a rate of 6 $ml.h^{-1}$ for 48 h.

The patient came with suspected surgical site infection on an emergency basis for a wound washout for the third time. Spinal anaesthesia was given in sitting position with 26 G Quincke needle with 0.5% hyperbaric bupivacaine 1 ml and fentanyl 20 µg as an adjuvant. Further course in the hospital was uneventful.

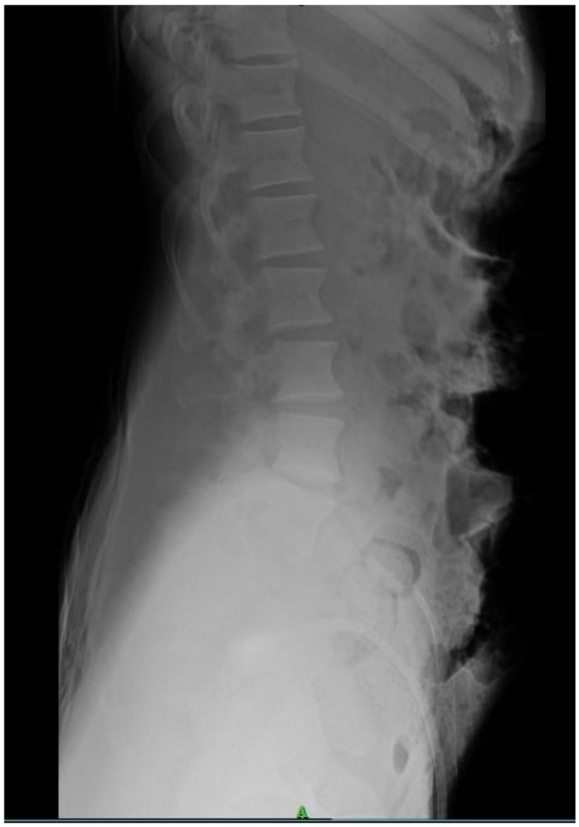

Fig. 6 Normal lumbar spine radiograph

Discussion

Pycnodysostosis (Toulouse-Lautrec syndrome) was described by Mareteux and Lamy in 1962. Incidence is 1.7 per 1 million births. The genetic defect is located on chromosome 1q21. This causes a mutation in cathepsin K, a lysosomal cysteine protease, which is highly expressed in osteoclasts, and causes osteosclerosis due to decreased bone resorption (Turan 2014; Puri et al. 2013; Kiran et al. 2008; Motyckova and Fisher 2002; Fleming et al. 2007; Mujawar et al. 2009). 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 (Bathi and Masur 2000; Hansen et al. 2018). Scoliosis, kyphosis and lumbar hyperlordosis are the typical features of the spine which can be seen in these patients (Beguiristain et al. 1995). Clinico-radiological diagnosis is of significance because of its resemblance with cleidocranial dysostosis and osteopetrosis (Puri et al. 2013). Clinical heterogeneity with its rarity challenges the prognosis in these patients (Bizaoui et al. 2019).

Literature describes the use of epidural anaesthesia for LSCS with pycnodysostosis (Hansen et al. 2018).

In our patient, planning for neuraxial anaesthesia was difficult with pregnancy, difficult airway, short stature, lack of radiological assessment of spine and associated fracture femur. Additionally, short stature made it difficult to decide the local anaesthetic dose for SA, and we wanted to avoid any risk of airway management later if spinal anaesthesia failed or partially worked. With concerns of a full stomach and difficult airway, an awake FOI with GA was planned. Conventional laryngoscopy/CMAC-assisted intubation was avoided because of severe retrognathia and to prevent any chance of cervical spine fractures. Magnesium sulphate given for neonatal neuroprotection did not add any significant concerns for recovery from neuromuscular block. Under USG guidance, nerve blocks played a beneficial role in postoperative analgesia.

After 2 weeks, she was planned for the surgical fixation of the fractured femur. A normal radiological assessment of the spine helped us to plan for a lumbar epidural catheterization. Epidural anaesthesia, tranexamic acid and maintenance of mean blood pressure of 60 mmHg helped in the prevention of significant blood loss. The patient had good postoperative pain relief with epidural analgesia. SA proved sufficient for the third surgical procedure of wound washout.

The myriad challenges in this case were precious and preterm pregnancy with fracture femur, anatomical and physiological changes of difficult airway, abnormal stature, risk of major blood loss, planning of analgesia, infection, risk of deep vein thrombosis due to recumbency, positioning and multiple surgeries in the same patient.

Conclusions

A case of pycnodysostosis with physiological and pathological considerations can present variedly to the anaesthesiologist in terms of surgical complications, the timing of presentation to operation theatre and for anaesthesia and analgesia. High-end armamentarium like fibre-optic bronchoscope and ultrasonography is demonstrated as useful aids in the anaesthetic management. Anaesthetic planning done contextually with each surgical setting helped in safe management of the patient. A careful interdepartmental planning, readiness and tailoring anaesthesia techniques bring out a better patient outcome.

Abbreviations

LSCS: Lower segment caesarean section; GA: General anaesthesia; FOI: Fibre-optic intubation; USG: Ultrasound; IV: Intravenous; SA: Spinal anaesthesia.

Acknowledgements
None.

Authors' contributions
All authors were involved in clinical management of case. RRK, conceptualization, primary draft, editing and reviewing and revision. SLG, editing and review and revision. PR, review and revision. VKZ, review and revision. PP, review and revision. The authors read and approved the final manuscript, that the requirements for authorship as stated earlier in this document have been met and that each author believes that the manuscript represents honest work.

Funding
None.

Availability of data and materials
Available on request with corresponding author.

Declarations

Ethics approval and consent to participate
The institutional review board of the Bangalore Baptist Hospital has approved the case report dated 22 March 2021, and written informed consent is taken from the participant.

Consent to publication
Written informed consent was obtained from the participant for publication purpose.

Competing interests
The authors declare that they have no competing interests.

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Received: 1 November 2021 Accepted: 23 September 2022
Published online: 08 October 2022

References

Bathi RJ, Masur VN (2000) Pyknodysostosis-a report of two cases with a brief review of the literature. Int J Oral Maxillofac Surg 29(6):439–442. https://doi.org/10.1016/s0901-5027(00)80076-4
Beguiristain JL, Arriola FJ, Leyes M (1995) Lumbar spine anomalies in a pycnodysostosis case. Eur Spine J 4(5):320–321. https://doi.org/10.1007/BF00301045 (PMID: 8581536)
Bizaoui V, Michot C, Baujat G, Amouroux C, Baron S, Capri Y et al (2019) Pycnodysostosis: natural history and management guidelines from 27 French cases and a literature review. Clin Genet 96(4):309–316. https://doi.org/10.1111/cge.13591 (Epub 2019 Jun 25 PMID: 31237352)
Fleming KW, Barest G, Sakai O (2007) Dental and facial bone abnormalities in pyknodysostosis: CT findings. Am J Neuroradiol 28(1):132–134
Hansen NS, Dalgaard KS, Jensen TB (2018) Anaesthetic considerations in a patient with pycnodysostosis undergoing caesarean delivery. Case Rep Anesthesiol. 2018:5675637. https://doi.org/10.1155/2018/5675637
Kiran S, Goel M, Singhal P, Goel M, Gupta R (2008) Pyknodysostosis: anaesthetic considerations – a case report. Internet J Anesthesiol 21(2):13
Motyckova G, Fisher DE (2002) Pycnodysostosis: role and regulation of cathepsin K in osteoclast function and human disease. Curr Mol Med 2(5):407–421. https://doi.org/10.2174/1566524023362401
Mujawar Q, Naganoor R, Patil H, Thobbi AN, Ukkali S, Malagi N (2009) Pycnodysostosis with unusual findings: a case report. Cases J 2(7):6544. https://doi.org/10.4076/1757-1626-2-6544
Puri R, Saxena A, Mittal A, Arshad Z, Dwivedi Y, Chand T et al (2013) Pycnodysostosis: an anaesthetic approach to this rare genetic disorder. Case Rep Anesthesiol. 2013:716756. https://doi.org/10.1155/2013/716756
Turan S (2014) Current research on pycnodysostosis. Intractable Rare Dis Res 3:91–93

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