

**Undiagnosed syndrome – A high clinical suspicion and meticulous anaesthesia is key to success**

<sup>1</sup>Dr. Pooja Upmanyoo, Junior Resident, Dept of anaesthesia, PGIMS, Rohtak

<sup>2</sup>Dr. Mamta Jain, Associate Professor, Dept of anaesthesia, PGIMS, Rohtak

<sup>3</sup>Dr. Anshul Mundra , Junior Resident, Dept of anaesthesia, PGIMS, Rohtak

<sup>4</sup>Dr. Dhruv Raheja, Junior Resident, Dept of anaesthesia, PGIMS, Rohtak

<sup>5</sup>Dr. Deepika Budhwar, Junior Resident, Dept of anaesthesia, PGIMS, Rohtak

<sup>6</sup>Dr. Aman Saini, Junior Resident, Dept of anaesthesia, PGIMS, Rohtak

**Corresponding Author:** Dr. Pooja Upmanyoo, Junior Resident, Dept of anaesthesia, PGIMS, Rohtak

**Citation this Article:** Dr. Pooja Upmanyoo, Dr. Mamta Jain, Dr. Anshul Mundra, Dr. Dhruv Raheja, Dr. Deepika Budhwar, Dr. Aman Saini, “Undiagnosed syndrome – A high clinical suspicion and meticulous anaesthesia is key to success”, IJMSIR- December - 2021, Vol – 6, Issue - 6, P. No. 114 – 116.

**Type of Publication:** Case Report

**Conflicts of Interest:** Nil

**Introduction**

Osteogenesis imperfecta is a rare autosomal disorder of connective tissue, associated with type 1 collagen abnormality caused by mutations in COL1A1 and COL1A2 genes.<sup>[1]</sup> Patients usually present for surgery due to recurrent fractures on trivial trauma. Anaesthetists often have to manage such patients without any proper diagnosis, only on the basis of high clinical suspicion. It affects bones, sclera and the inner ear.<sup>[1]</sup> Various anaesthetic concerns are highly fragile bones, thoracic deformities leading to decrease in vital capacity and chest wall compliance, a tendency to develop malignant or non malignant hyperthermia and platelet dysfunction.<sup>[3-5]</sup> Hereby, we describe the successful anaesthetic management of an 11 year old child with history of frequent fractures and blue sclera planned for fracture olecranon surgery.

**Case Report**

An 11 year, 125 cm tall old boy, weighing 35 kgs, presented with fracture olecranon left forearm planned

for open reduction and internal fixation under general anaesthesia. Patient had history of multiple fractures of upper limbs in the past for which he got operated once under GA, uneventfully but no records available. In family history patient’s mother too had blue sclera but no history of frequent fracture and no history of GA for any reason. Patient had laxity of joints, blue sclera and thin skin. On general examination he was alert, conscious and afebrile. His pulse and blood pressure were in normal limits. Respiratory system revealed barrel shaped chest with equal entry bilaterally. Heart sounds were muffled. Airway examination was within normal limit with Mallampatti class I. Routine haematological investigations were in normal limits. Electrocardiography and X-ray chest were in normal limits. We referred the patient to cardiologist for echocardiography to rule out any valvular cardiac anomaly. Echocardiography revealed trace TR. Patient was taken for surgery with all risks explained and full preparation. Anaesthesia machine was prepared by

flushing at high fresh gas flow for 15-20 minutes. In the operation theatre patient was carefully placed in supine position. Routine monitors (ECG, SpO<sub>2</sub>, skin temperature probe) were applied and blood pressure was taken manually. Intravenous line secured in right hand with 20 gauge cannula. Agents triggering malignant hyperthermia were avoided. Patient was preoxygenated with 100% oxygen for 3 minutes. Anaesthesia was induced with propofol (2 mg/kg) and fentanyl (2 µg/kg). Check ventilation was done and injection vecuronium 4 mg was administered. Intermittent positive pressure ventilation done for 3 minutes. Laryngoscopy was performed in neutral position and patient intubated with ETT of 6.5 mm ID. Patient was put on volume controlled mode and End tidal CO<sub>2</sub> monitoring was in place throughout the peri operative period. Patient was made in right lateral position with utmost care in the form of appropriate padding on pressure areas with cotton pads and preformed cushions.

Anaesthesia was maintained with total intravenous anaesthesia (TIVA) using propofol ( 2 mg/kg/hr) and fentanyl (1 µg/kg/hr ) and intermittent dose of vecuronium. Ventilation was continued with a mixture of oxygen in air (50:50) using a closed anaesthetic circuit, with a tidal volume of 8 ml/ kg and respiratory rate of 14/minute. Surgery was completed in 2 hours. Vitals were stable throughout the procedure. At the end of the surgery patient was made supine carefully and reversal was done using 100% oxygen with glycopyrrolate 0.4 mg and neostigmine 2 mg. Patient extubated uneventfully in a deeper plane after resumption of spontaneous respiration to prevent return of excessive muscle tone. After monitoring in recovery,

patient was transferred back to ward. Patient was subsequently discharged on 5<sup>th</sup> post-operative day.

### Discussion

Osteogenesis imperfecta is of two types: congenita form, with a high mortality rate and the tarda form, which is associated with normal life expectancy.<sup>[2]</sup> These patients possess several challenges for the anaesthesiologist. Due to hypermobile joints, difficult airway must always be anticipated in these patients.<sup>[6]</sup> Therefore, we also prepared a difficult airway cart before induction. Hyperextension of neck during laryngoscopy can cause cervical spine fracture or atlanto-occipital dislocation so we preferred laryngoscopy in neutral position. Inhalational vaporizers were removed before taking the patient. We did not use any inhalation agents as isoflurane, sevoflurane to avoid development of malignant hyperthermia. We also avoided suxamethonium for the same reason, also suxamethonium induced fasciculations may cause fractures in these patients.<sup>[7]</sup> We continuously monitored the skin temperature. An automated blood pressure cuff may overinflate and can result in fracture therefore we used manual sphygmomanometer.<sup>[8]</sup> The bleeding may occur despite normal coagulation profile so due precautions were taken regarding any unexpected bleeding and adequate blood and blood products were kept ready.<sup>[5,9]</sup> Utmost care was taken during positioning of patient to prevent any fractures and dislocations. We conclude that TIVA with propofol and fentanyl provided haemodynamic stability and unchanged temperature throughout the procedure.

### Conclusion

Undiagnosed syndromic patient pose a significant challenge to anaesthesiologist owing to risk of difficult

airway and associated multiple co morbidities. Hence, regional anaesthetic technique should be preferred wherever permissible and in case general anaesthesia is required, avoid malignant hyperthermia triggering agents and maintain anaesthesia using TIVA.

### References

1. Marini JC. Osteogenesis imperfecta: managing brittle bones N Eng J Med. 1998;339(14):986–987.
2. Stynowick GA, Tobias JD. Perioperative care of the patient with osteogenesis imperfecta. Orthopedics. 2007;30(12):1043-1049.
3. Porsberg P, Astrup G, Bendixen D, et al. Osteogenesis imperfecta and malignant hyperthermia. Is there a relationship? Anaesthesia. 1996;51(9):863–865.
4. Baines D. Total intravenous anaesthesia for patients with osteogenesis imperfecta. Pediatr Anaesth 1995; 5: 144.
5. Garg M, Jain M, Gupta A. Anaesthetic management of a case of osteogenesis imperfecta with urinary bladder stone: a case report. Indian J Anaesth. 2009;53(1):68-70.
6. Malde AD, Jagtap SR, Pantvaidya SH, Kenkare JS. Osteogenesis imperfecta: anaesthetic management of a patient for abdominal hysterectomy (a case report). Indian J Anaesth. 1993;41 : 203–206.
7. Kostopanagiotou G, Coussi T, Tsaroucha N, Voros D. Anaesthesia using a laryngeal mask airway in a patient with Osteogenesis Imperfecta Anaesthesia 2000;55:489–518
8. Bhandari G, Shahi KS, Bhadoria P, et al. Osteogenesis imperfecta: no place for an imperfect anaesthesiologist. Indian J Anaesth. 2008;52(5):577.

9. Wong RS, Follis FM, shively BK, Wernly JA. Osteo-genesis imperfecta and cardiovascular diseases. Ann Thorac Surg 1995;60:1439–43.

### Legend Figure

