CASE REPORT

Ellis-Van Creveld syndrome anaesthetic approach

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Abstract

Ellis–van Creveld (EVC) syndrome, also known as Chondroectodermal dysplasia, is a rare autosomal recessive disorder. A preoperative meticulous systemic wise assessment of patient guides in early optimization prior to surgery. Postoperative pain must be tackled conscientiously, with preparation to circumvent any adverse cardio respiratory events. Following is a case report of 13-year-old female patient diagnosed case of EVC syndrome posted for lower limb correction surgery. We have described the anaesthestic approach, issues faced during induction and how it can be successfully tackled for a better intraoperative and postoperative care of patient.

Keywords: Ellis van Creveld syndrome, Difficult Epidural, Patchy Subarachnoid Block

Introduction

Ellis-van Creveld (EVC) syndrome is a rare autosomal recessive disorder. Approximately 300 cases have been reported, with higher incidence in Amish Communities in the United States of America [1]. Research claims that mutations affect signalling pathways at cellular level and hence EVC have multi-organ involvement. EVC has two components: (1) chondrodysplasias, which are characterised by disorders of bone maturation, and (2) ectodermal dysplasia which involves ectodermal derived tissues such as teeth, nails, hair and skin [1-2].

Case Report

A 13 year old female patient presented with chief complaints of bilateral knee pain, walking difficulty and limb deformity aggravated since 2 years. Her weight was 50 kg, height 130 cm (disproportionate short stature). She was a diagnosed case of EVC syndrome. On clinical examination she had post axial polydactyly with hypoplastic finger nails, genuvalgus and gait abnormalities. On airway examination, mouth opening and neck movements were adequate however she had conical teeth, malocclusion with musculofibrous frenula as in Figure 1. On auscultation no murmurs were detected. On spine examination she had lordosis of lumbar spine. Difficult venous access was noted. Echocardiography showed ejection fraction of 60-65% with trace tricuspid regurgitation. All the remaining lab investigations were normal.

After taking written informed consent from parents and confirming nil by mouth status, patient was shifted to the operation theatre and standard monitors were attached. Adequate care was taken during patient positioning. Our initial plan was Subarachnoid Block (SAB) with Epidural Anaesthesia (EA) considering her difficult airway status. For combined spinal EA, the patient was placed in sitting position. Initial attempts to secure epidural via median approach failed. Subsequently, paramedian approach at L3-L4 was attempted with the

loss of resistance technique. Despite successful identification of epidural space, catheter could not be advanced further after a certain limit (Figure 2a). Hence, procedure was abandoned. Hence, we decided to proceed with SAB using 26G Quincke needle at L2-L3 space. After confirmation of clear flow of cerebrospinal fluid, 2.8 ml of 0.5% heavy bupivacaine with 5mcg of dexmedetomidine was administered (Figure 2b). After waiting for 25 minutes patient had only patchy block with Bromage scale 3. As a result, we decided to go ahead with General Anaesthesia (GA). Following pre-oxygenation for 3 minutes and pre-medication with Intravenous (IV) glycopyrrolate 0.2 mg, midazolam 1mg and ondansetron 4 mg, patient was induced slowly with IV fentanyl 80 mcg and propofol 100 mg. After confirmation of bag and mask ventilation, 10 mg of cisatracurium was given and using video laryngoscope (Figure 3), endotracheal tube of 6 mm internal diameter tube was inserted smoothly. Pain was managed with IV 1gm paracetamol and morphine 2 mg boluses. Patient underwent peroneal nerve decompression with tibia wedge osteotomy (Figure 4). The mean duration of surgery was 240 minutes. Total blood loss was 100 ml. At the end of surgery with return of spontaneous breaths reversed with IV Glycopyrrolate 0.01 mg/kg with Neostigmine 0.05 mg/kg, smooth extubation was performed. Patient was shifted to intensive care unit for postoperative monitoring.

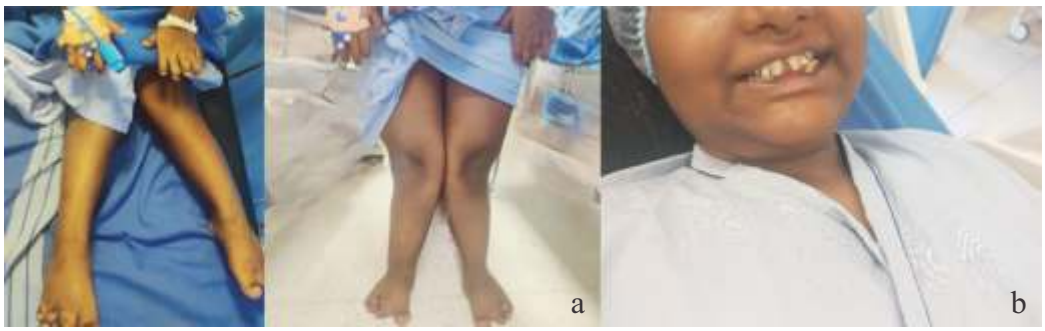

Figure 1a: Genuvalgum
Figure 1b: Conical hypoplastic teeth

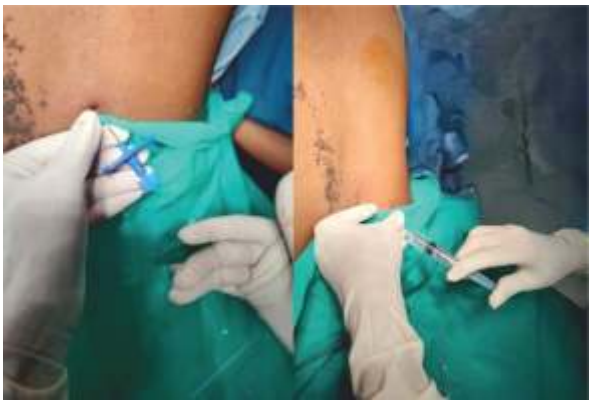

Figure 2a: Epidural catheter obstructed
Figure 2b: Subarachnoid block administration

Figure 3: Video laryngoscopic view

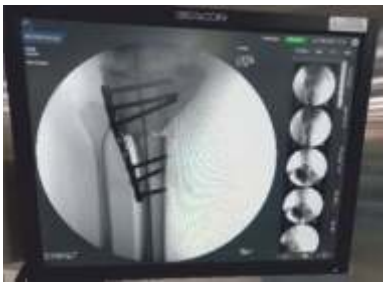

Figure 4: Surgical correction

Discussion

EVC being an autosomal recessive trait can be acquired from parents with consanguineous marriage or other affected members. In our case there was history of parental consanguinity. There is 50% mortality in EVC syndrome, encountered in patients with cardio respiratory abnormalities [3]. Cardiac anomalies include atrial septal defect, ventricular septal defect, and hypoplastic left heart syndrome [4-5]. Our pre-operative cardiac echocardiography were within acceptable limits.

Hepato-renal anomalies reported were renal medullary dysplasia, nephrotic syndrome and refractory cirrhosis [5]. Our patient was posted for genu valgus corrective surgery. IV access can be difficult in such patients secondary to excessive lax skin and subcutaneous tissue.

Our initial plan was SAB with EA keeping in mind the respiratory and cardiac status of the patient, her difficult airway status, lower limb surgery, and better postoperative pain management. Our patient had spine deformity, hence epidural space identification was difficult. We managed to identify epidural space via para median approach but catheter could not be advanced further after a certain limit. Hopman *et al.* described in their case report wherein after initial identification of epidural space, difficulty was encountered in catheter placement. Such obstruction was probably due to spine deformities such as smaller pedicels and narrow bony canal [6]. We later proceeded to perform SAB but despite waiting for 20 min, it was an inadequate block. Kaur *et al.* have highlighted in their case report about the possible reasons of failed spinal anaesthesia in patients with spine deformity like anatomical defect, spinal canal stenosis or sclerosis which prevents drug spread or

defective drug [7]. Fernández-Meré *et al.* also reported a case of EVC syndrome posted for caesarean section, in whom due to failed SAB they induced patient with GA [8]. Nayak *et al.* have highlighted in their case study that underlying spine deformity can lead to defective spread of anaesthetic solution while performing regional block which may eventually lead to failure of regional anaesthesia altogether [9].

We proceeded with GA and secured airway with ETT. Any deformity comprising maxilla, mandible and frenulum leads to difficult mask ventilation. Dental anomalies can lead to tooth dislodgment during intubation. In view of above complications difficult airway cart was kept ready. Intubation was performed smoothly using video laryngoscopy. Our choice was cisatracurium as it is an intermediate acting, non-depolarizing neuromuscular blocking drug. It is three times more potent than atracurium. It undergoes spontaneous degradation and has lesser dose related histamine release, with cardiovascular stability in both healthy patients and those with coronary artery disease. Recovery rate was unaffected by age, renal or liver disease. Our choice has been justified by Abeles *et al.* who observed safety in using midazolam, fentanyl, dexmedetomidine, and cisatracurium for intubation in a patient scheduled for cardiovascular surgery [10]. Possibility of bronchial cartilage hypoplasia in EVC patient has been reported so we anticipated post intubation airway collapse. As a result, we abstained from high airway pressures to lessen barotraumas occurrence. Care should be taken in patient positioning due to existing contractures in EVC patients. Pain was managed using IV paracetamol, opioids like fentanyl, later morphine boluses were given. As a result, patient was relatively pain free in the postoperative period.

In our experience the main aim of anaesthetic management is extensive airway examination, check for underlying limb and spine deformities, followed by early assessment and optimisation of multisystem involvement. In addition, excellent pain management and prevention of further cardio respiratory events are other equally important aspects in anaesthetic management [9-10].

Conclusion

In view of the above knowledge we would like to emphasise the importance of multidisciplinary team approach with alternative anaesthesia planning for betterment and smooth course of patient in intraoperative and postoperative period.

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