

Bronchospasm in a case of Ellis van Creveld syndrome in a patient posted for corrective osteotomy & elizarove surgery

M Prakash, A Swain, S mishra, A Badhe

Citation

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Abstract

Ellis-van Creveld syndrome (chondroectodermal or mesoectodermal dysplasia) is an autosomal recessive disorder. Initial reported as tetrad of clinical manifestations, including chondrodysplasia, postaxial polydactyly, ectodermal dysplasia, and congenital heart disease (CHD). Other involvement may occur in organs of endodermal origin, such as the pulmonary, renal, gastrointestinal, and central nervous systems. A known case of Ellis van Creveld syndrome was scheduled for elective corrective osteotomy and elizarove surgery. Here with we describe the clinical features of a patient and anaesthetic management of our patient

INTRODUCTION

Ellis-Van Creveld Syndrome or chondroectodermal dysplasia is a form of short-limbed dwarfism. The name chondroectodermal is used because it affects the skeleton (chondro) and the skin (ectoderm).¹The syndrome was first described by Ellis and Van Creveld² in 1940. Reported incidence is one in 1500000.¹The highest frequency of Ellis–van Creveld syndrome is seen in one particular inbred population, the Old Order Amish community in Lancaster County, Pennsylvania, where the largest pedigree has been described (52 cases in 30 sib ships).³ Its incidence in India is very rare. Literature search from publication of first case in 1940 to date revealed only five case reports from this region.

CASE REPORT

A 12 year old child weighing 25 kg was posted for corrective osteotomy & elizarove surgery of tibia. Pre operative evaluation of the patient previous day showed spiky hair, high arched palate, syndactily, polydactyly, loss of lateral condyle of tibia, widened carpal bones, dwarfism and genu valgum.

Figure 1

Figure 1 (Postaxial polydactyl)



Figure 2

Figure 2 (Knock knees or Genu-valgum)



Cardiac anomalies were ruled out by doing an echocardiogram. Lab values were Hb 11 gm%, blood sugar 101 mg/dl (random), blood urea 19 mg/dl, serum creatinine 0.9 mg/dl, serum sodium 140mEq/l, potassium 4.13meq/l. The patient was partially edentulous with cleft palate and. Birth history showed normal delivery. There was no birth asphyxia or delayed milestones. Preoperatively patient was explained about the advantages of regional anaesthesia. But the patient accepted epidural catheter insertion only under general anaesthesia. The patient was given anaesthetic fitness under ASA class I. He was premedicated with Tab. Famotidine 10 mg previous day night and Tab. Famotidine 10mg and Tab. metoclopramide 5 mg on the morning of surgery. Once the patient was shifted to Operation theatre, 20 gauge I.V. line was secured on the left hand. Later ECG, NIBP and SpO₂ monitors were connected. Base line readings on the table were pulse rate 68/min, blood pressure 100/60 mmHg, room air SpO₂ 100%. 40 µg of fentanyl intravenously and IV 0.5mg of midazolam premedication received on table. The patient was preoxygenated with 100% oxygen for 3 minutes and induced with inj. Thiopentone sodium 150 mg and paralysed with inj. Vecuronium bromide 3.2 mg intravenously. The patient was ventilated with mask with N₂O and O₂ (4 and 2 liters/min respectively) + isoflurane (1%) for 3 minutes. Intubated with no.6 cuffed endotracheal tube. Bilateral air entry was confirmed by auscultation and endotracheal tube was firmly secured. Later the patient was positioned in left lateral position and under aseptic conditions 20 gauge epidural catheter was inserted at L4 – L5 space to an intra epidural length of 4cm. Epidural catheter position at epidural space was confirmed by a test dose consisting of 3cc of 2% lignocaine with 15 µg of adrenaline. Patient was positioned supine and 6cc of 2%

lignocaine was given through epidural catheter. Anaesthesia was maintained with N₂O and O₂ (2 and 1 liters/min respectively) + isoflurane (0.2 - 1%) + IPPV. Analgesia was maintained with 3cc boluses of 0.5% bupivacaine given through the epidural catheter every 60 minutes interval. The patient needed a total of 5 doses as the procedure was lasted for 6 hrs. Muscle relaxation was maintained with inj. vecuronium bromide top ups (0.4mg) as and when desired as determined by EtCO₂. Throughout procedure the patient was monitored with ECG, SpO₂, NIBP (every 5 minutes interval), EtCO₂, end tidal agent monitoring (N₂O, O₂ and isoflurane), urine output and airway pressure. Total blood loss during the procedure was 300ml. Totally 1000ml of crystalloid 500ml of colloid and 200ml of packed cell was given. Total Urine output was 600ml. Last dose of bupivacaine was mixed with 3mg of morphine.

During the procedure two episodes of increase in airway pressure (from 16 cmH₂O to 30 cm of H₂O) was observed and on auscultation bilateral wheeze was appreciated along with drop in oxygen saturation to 92%. Bronchospasm was managed with nebulisation salbutamol. Inj. Hydrocortisone 50mg and inj. theophylline (at first episode) and by increasing isoflurane concentration (at second episode). Once procedure completed the patient was reversed with neostigmine (1.5mg) and atropine 0.9mg and extubated on table. Postoperatively analgesia was supplemented with the epidural morphine 3mg bolus as and when the patient demanded for 24 hrs (patient demanded 2 boluses). Post operative vitals were stable.

DISCUSSION

Richard W.B. Ellis of Edinburgh and Simon Van Creveld of Amsterdam first described Ellis-van Creveld (EVC) syndrome. They met in a train compartment while travelling to a paediatrics conference in England in the late 1930s, and discovered that each had a patient with the syndrome. Disproportionate dwarfism, postaxial polydactyly, ectodermal dysplasia, a small chest, and a high frequency of congenital heart defects (Table-1) characterize this autosomal recessive syndrome⁴. Pathophysiology is unknown. Recently two EVC genes were identified. This may through some light on pathophysiology in future. Morbidity and mortality is related to the thoracic dysplasia, respiratory insufficiency and cardiac anomalies. This leads to death in infancy in 50% of patients⁵. Frequency is equal in both sexes.

Figure 3

Table 1

Ellis-van Creveld Syndrome (chondroectodermal dysplasia)	
Airway	Micrognathia, cleft lip or palate. May have a short trachea Alveolar ridge hypoplasia. Multiple maxillary and mandibular frenula attach to the alveolar ridge. Short upper lip ("partial harelip"), bound by frenula to alveolar ridge (lip tie). Frenula attach to the alveolar ridge. A fusion of the anterior portion of the upper lip to the maxillary gingival margin. Teeth may be prematurely erupted at birth or exfoliate prematurely. Neonatal teeth, hypoplastic or missing teeth.
Respiratory	Narrow Long, thorax with short ribs. Restrictive lung disease and pulmonary hypoplasia. May have Bronchial cartilage hypoplasia with lobar emphysema. Recurrent pneumonia
Cardiovascular	Heart defects occur in 50-60% of patients. Most common anomaly is a common atrium (40%). Other anomalies include AV canal Defect, VSD, ASD and PDA.
Genitourinary	hypospadias, epispadias, cryptorchidism, vulvar atresia, nephrocalcinosis, renal agenesis, and megaureters.
Central Nervous	CNS anomalies or mental retardation are present, Dandy-walker malformation
Musculoskeletal	Low-set shoulders, a narrow thorax frequently leading to respiratory difficulties, knock knees, (Genu valgum), lumbar lordosis, broad hands and feet, and sausage-shaped finger

The clinical features that are suggestive of Ellis van creveld syndrome in our patient are spiky hair, high arched pallet, syndactily, polydactyly, loss of lateral condyle in the tibia, widened carpal bones, dwarfism, partially edentulous & genu valgum. There are no cardiac anomalies which were ruled out by doing echocardiogram of heart. Literature says that cardiac anomalies may be present only in 50% of cases¹. Total height of our patient is 104 cm. For a child who is 12 yrs old this height is very less.

As the patient was anxious and as it was a prolonged procedure we had chosen for a combined procedure i.e. combined epidural and general anaesthesia. During the procedure twice we had encountered bronchospasm. It might be due to patients' intrinsic problem because of the

syndrome (small chest, recurrent infection, and restrictive lung disease) which was not obvious when the patient was conscious, but which was encountered during anaesthesia.

CONCLUSION

Though cardiovascular problem which is present in 50-60% of cases with Ellis-Van Creveld Syndrome is the major concert of anaesthetic management (which is not so in this case), respiratory problems like small narrow chest , restrictive lung disease, recurrent infection, pneumonia, lobar emphysema are also major part of anaesthetic management. Preoperative evaluation for Ellis-Van Creveld Syndrome should also include Chest X-ray, CT-scan chest, and pulmonary function test to rule out restrictive lungs disease.

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Author Information

M.V.S.Satya Prakash, MBBS, DA, DNB

Senior resident, Department of Anaesthesiology and Critical Care JIPMER

Anwesa Swain, MBBS, MD

Senior resident, Department of Anaesthesiology and Critical Care JIPMER

Sandeep kumar mishra, MBBS, MD

Assistant professor, Department of Anaesthesiology and Critical Care JIPMER Pondicherry-6 India

AS Badhe, MD, DA

Professor and HOD Department of Anaesthesiology and Critical Care JIPMER Pondicherry, India