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Anaesthetic management of patient with Ellis Van Creveld syndrome

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Abstract

A known case of Ellis-Van Creveld Syndrome was scheduled for emergency repair of obstructed paraumbilical hernia. We describe the anaesthetic management of the case with special reference to the classic physical and physiological manifestations of this syndrome present in 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).¹ The syndrome was first described by Ellis and Van Creveld² in 1940 and this disease was found mainly among the Amish group of population chiefly in Pennsylvania, Ohio and Indiana. The disorder is characterized by anomalies of the hands and ectodermal dysplasia involving the nails and teeth. Reported incidence is one in 1,500,000 live births.¹ Incidence in Pakistan and 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 36 year old female, known case of Ellis-Van Creveld Syndrome, presented in the hospital emergency room with vomiting, abdominal swelling and pain. A diagnosis of obstructed paraumbilical hernia was made and the patient was scheduled for emergency repair of the obstructed hernia.

On preoperative evaluation, patient had short stature, short limbs in relation to the trunk, polydactyly of the hands (Figure 1), protuberant abdomen, narrow chest and moderate lumbar lordosis. She was morbidly obese with a



Figure 1. Polydactyly in hand. Ulnar artery cannulated for invasive arterial pressure monitoring.

body mass index (BMI) of 47.87. There was no abnormality of cervical spine and airway examination was unremarkable. Blood pressure was 160/100 mmHg and heart rate 95-100/min. The white blood cell count was 23,000/cmm and baseline arterial blood gases showed a PaO₂ of 65 mm Hg and SaO₂ of 92% on room air.

An echocardiogram done one year back showed a moderately dilated right atrium, mild right ventricular hypertrophy, large atrial septal defect (secundum type) with left to right shunt and severe pulmonary stenosis. The left ventricular systolic function was normal. The patient was evaluated by cardiologists and placed in high cardiac risk category for any kind of surgical intervention under general anaesthesia.

Supplemental oxygen was started via face mask preoperatively. Aspiration prophylaxis (Syrup Sodium citrate 0.3 M 30 ml, Inj. Ranitidine 50 mg and Inj.

Metoclopramide 10 mg intravenously) was given an hour prior to induction of anaesthesia considering the high risk of regurgitation and aspiration due to obstructed hernia and morbid obesity.

General anaesthesia with endotracheal intubation and invasive cardiovascular monitoring was planned. Any neuraxial block for pain management was not considered due to thoracic and lumbar skeletal deformity. She was labeled as American society of anesthesiologist (ASA) class IVE.

Pre-induction, invasive arterial monitoring was initiated by cannulation of left ulnar artery (Figure 1) because radial pulse was not easily palpable. After preoxygenation, a modified rapid sequence induction was done with fentanyl 150 µg, etomidate 14 mg and succinylcholine 100 mg. The trachea was intubated with endotracheal tube (ETT) size 7.0 mm. The right internal jugular vein was cannulated for monitoring of central venous pressure and fluids. A Foleys catheter was placed for monitoring of urine output.

General anaesthesia with pressure control ventilation was maintained with isoflurane 1.5 -2 % and 50% oxygen in air. Neuromuscular blockade with boluses of atracurium and systemic analgesia with fentanyl was continued during the maintenance period. Glyceryl trinitrate (GTN) infusion was used to control blood pressure. The patient remained haemodynamically stable throughout the surgical procedure which lasted for three hours and intraoperative blood loss was insignificant. Postoperatively, she was admitted to the intensive care unit (ICU) for mechanical ventilation and invasive cardiovascular monitoring due to presence of several risk factors for postoperative respiratory insufficiency as well as early detection and rapid control of cardiovascular instability.

She was extubated uneventfully the next day and kept under observation for 24 hours. She was subsequently transferred to the special care unit and discharged from hospital three days later.

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 traveling 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 characterize this autosomal recessive syndrome³, which has an increased incidence among persons of old order Amish descent.

Pathophysiology is unknown; however, recent

identification of the EVC gene should lead to a better understanding. Histopathologic examination of fetuses with Ellis-van Creveld syndrome revealed that the cartilage of long bones showed chondrocyte disorganization in the physal growth zone.

In the general population, the frequency is 1 case per 60,000 live births. Among persons from the Old Order Amish, the incidence is estimated at 5 cases per 1000 live births. The frequency of carriers in this population may be as high as 13%. Morbidity and mortality is related to thoracic dysplasia, respiratory insufficiency and cardiac anomalies. This leads to 50% deaths in infancy. There is no sex preponderance. In our patient significant, cardiac anomalies, thoracic dysplasia and morbid obesity were present.

Family history may include parental consanguinity or previously affected siblings or family members. Neonatal history may reveal small size at birth, slow growth, and skeletal anomalies. Natal teeth may be present. Heart disease may be manifested as failure to thrive, cyanosis, shortness of breath, cardiac murmur, or other signs suggestive of heart failure. Developmentally, most patients have had intelligence in the normal range.

Diagnosis rests solely upon skeletal anomalies. Expected findings include relative shortening of the distal and middle segment of the limbs which is most prominent in the hands, where the distal and middle phalanges are shorter than the proximal phalanx. Polydactyly⁴ is common on ulnar side, as it was present in our patient while valgus deformity of the knee and fibula disproportionately smaller than the tibia. Chest radiograph, ECG and echocardiogram are required to evaluate cardiac anomalies. Foetoscopy and ultrasonography may be performed for prenatal diagnosis. Occasionally E.V.C. has to be differentiated from asphyxiating thoracic dysplasia or Asphyxiating thoracic dystrophy⁶ (Jeune syndrome) and short limb polydactyly syndrome type 3 because of overlapping features.

The primary goal of medical care is supportive and care should be taken for respiratory distress, recurrent respiratory infections, cardiac failure and dental care in childhood. Prevention of caries with dietary counseling, plaque control, and oral hygiene instruction should be given to parents. The surgical procedures are usually orthopaedic to correct polydactyly and valgus deformity. Final adult height is 43-60 inches. Usually, some limitation of hand function exists, such as inability to form a clenched fist. Dental problems are frequent.

The primary goal of anaesthetic management is thorough evaluation of skeletal deformities, airway abnormalities, restrictive ventilatory defect and cardiac

anomalies. The magnitude of decompensation should be assessed and optimized if possible. The aim of intraoperative management is to gain rapid control of airway, prevention of aspiration, barotrauma, swings of blood pressure, myocardial depression, worsening of pulmonary hypertension and left to right shunt, reversal of shunt and cardiac failure. Ventilatory management is directed towards maintaining stable airway pressures, normoxia and normocarbia. Postoperative management should constitute adequate analgesia and prevention of adverse cardio-respiratory events.

Conclusion

Our patient showed many of the characteristic changes described in literature. No case reports regarding anaesthetic management of these patients have been found. The principal goal of anaesthetic management in these patients is to maintain cardiorespiratory stability, with use

of invasive haemodynamic monitoring and ventilatory support, continued into the postoperative period.

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