Case Report

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Anaesthetic management of patient with Ellis Van Creveld Syndrome

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ABSTRACT

Ellis van Creveld syndrome (EVC syndrome) also known as chondroectodermal or mesoectodermal dysplasia was first described by Richard W.B. Ellisand Simon Van Creveld in 1940. The name chondroectodermal is used because it affects the skeleton (chondro) and skin (ectoderm). In general population the incidence is reported as 7 per 1,000,000 live births. Due to multisystem involvement anaesthesia care is challenging during intraoperative as well as post-operative period. Here we describe the perioperative care of a 14 year old female with EVC syndrome who underwent corrective osteotomy of tibia for genu valgum. Patient was given anaesthesia fitness under American Society of Anesthesiologists as grade 1. We planned to give subarachnoid block to the patient as she had no anomalies involving any other system of body except skeletal system, moreover our patient had normal lumbosacral spine study but thoracic spine scoliosis. Perioperative period was without any adverse events. Postoperative management constitutes adequate analgesia and prevention of adverse cardiorespiratory events.

Key words: Ellis van Creveld syndrome, genu valgum

INTRODUCTION

Ellis van Creveld syndrome (EVC syndrome) also known as chondroectodermal or mesoectodermal dysplasia was first described by Richard W. B. Ellisand Simon Van Creveld in 1940[1].

The name chondroectodermal is used because it affects the skeleton (chondro) and skin (ectoderm)[2]. Organs of endodermalorigin mayshows involment like pulmonary, renal, gastrointestinal, hematological and central nervous system. Due to multisystem involvement anaesthesia care is challenging during intraoperative as well as post-operative period. Here we describe the perioperative care of a 14 year old female with EVC syndrome who underwent corrective osteotomy of tibia for genu valgum.

CASE REPORT

A 14 year old female with EVC syndrome was posted for corrective osteotomy of tibia for genu valgum. She weighed 26 kgs and her height was 126 cm. She had short stature,short limbs in relation to trunk,polydactyly of right hand and genu valgum. Finger and toe nails were small and brittle. She had knock kneeswith pectus excavatum. Oral cavity examination showed absent incisors and rest of teeth were natal teeth but none of the tooth was loose. Airway examination showed normal mouth opening, normal range of movement of neck and Mallampati class I. On examination of spine, thoracic scoliosis was seen. She attained milestones appropriate with age, had normal intelligence and attained menarche at the age of thirteen. Cardiopulmonary examination showed normal heart soundswith no murmur and air entry was equal on both sideswith no added sounds. Haemogram, blood sugar, liver function, renal function tests, coagulation profile and serum electrolyteswere within the normal range. Electrocardiogram, ultra-sonogram of abdomen and pelvis showed normal study. Echocardiography showed septal aneurysm bulging into right atrium with no clot. Left

How to cite this article: How to site the article: Lalwani J, Patel M, Shah PJ. Anaesthetic management of patient with Ellis Van Creveld Syndrome Central Journal of ISA 2017;1(2):78-80.

ventricular systolic function was normal. X-ray spine showed thoracic scoliosis with normal cervical, thoracic spine and lumbar spine.

Preoperatively patient was explained about the advantages of regional anaesthesia and informed consent was taken for the same. We planned to give subarachnoid block to the patient. Patient was given anaesthesia fitness under American Society of Anaesthesiologists as grade 1. She was premedicated with tablet Famotidine 10 mg previous night and on the morning of surgery along with tab metoclopramide 5 mg, 30 mins prior to surgery. Once the patient was shifted to operation theatre, 20 gauge intravenous line was secured on the left hand and ringer lactate was started at the rate of 10 ml/kg/hr and monitor (Philips Multi ParaPMS Intelliuve MP 30 Monitor) was attached for non-invasive Blood Pressure (BP), electrocardiogram, oxygen saturation (SPO2).Her baseline reading of vital parameters was recorded as BP 126/86, Heart Rate (HR) 86/min, SPO2=100% on air). Sub arachnoid block was given in sitting position under all aseptic precautions with 25 gauge Quincke's needle at L3-L4 interspace after infiltration of 2ml of 2% lignocaine in skin and subcutaneous plane, 2.5 ml of 0.5% bupivacaine heavy was injected. The patient was immediately turned supine and height of the block was assessed. Maximum level of the block was upto T12. Urinary catheter was inserted. Patient remained haemodynamically stable throughout intra-operative period which lasted for 2 hours, intra operative blood loss was insignificant. Monitoring of all vital parameters such as ECG, NIBP, SPO2 was done at 5 mins interval throughout the intra operative period.After the end of surgery she was transferred to the Intensive Care Unit (ICU) for monitoring and then to the postoperative ward after 24 hrs from where she was discharged after five days. Postoperative analgesia was given as i.v. fentanyl

DISCUSSION

Ellis van Creveld syndrome is an inherited autosomal recessive disorder, characterized by clinical tetrad of chondrodystrophy or disproportionate dwarfism, post axial polydactyly, ectodermal dysplasia (mainly affecting teeth, nail and hair) and congenital heart disease[1,3]. Polydactyly is common on ulnar side, which was present in our patient[4].

In general population the incidence is reported as 7 per 1,000,000 live births with an increased incidence of 5 per 1000 live births in old Amish population of Pennsylvania with no sex prelidiction.[3,5]. Consanguinity is implicated in approximately one-third of the cases[5]. It results from amutation in one of Ellis-van-Creveld genes (EVC1 and EVC2) located on short arm of chromosome 4(4p16) which is found in approximately two-third of EVC cases[6–8]. Morbidity and mortality is related to thoracic dysphasia, respiratory insufficiency and cardiac anomaly. This leads to 50 % deaths in infancy[9]. But fortunately our patient had normal cardiac function as evaluated preoperatively by echocardiography.

Neonatal history may reveal small size at birth, slow growth and skeletal anomalies. Natal teeth may be present, heart disease may manifest as failure to thrive, shortness of breath, murmur or other signs suggestive of heart failure. Most patients have intelligence in the normal range[8]. So the primary goal of anaesthetic management in such cases is pre anaesthetic evaluation of skeletal deformities, airway abnormalities, others such as renal, hepatic, pulmonary and cardiac anomalies .Another consideration in E-V-C syndrome that may impact anaesthesia care include renal and hepatic involvement[10,11]. Preoperative evaluation of renal function including serum electrolyte, Blood Urea Nitrogen (BUN), serum creatinine and urinary analysis appears essential which was normal in our patient.Liver function tests were also within the normal range.

Our patient had no such anomalies involving any other system of body except skeletal system, moreover our patient had normal lumbosacral spine study and thoracic spine scoliosis. So we planned regional anaesthesia (subarachnoid block) for corrective osteotomyof tibia to avoid systemic complication of general anaesthesia. We did not encounter any problem in performing subarachnoid block. Postoperative management constitutes adequate analgesia and prevention of adverse cardiorespiratory events.

CONCLUSION

Patients with Ellis van Creveld syndrome can be effectively managed provided careful consideration is given for assessment of organ functions; regional anaesthesia can be safely administered with successful outcomes.

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