Brief Communication

Anaesthetising an infant with acrocallosal syndrome: An unusual case

INTRODUCTION

Acrocallosal syndrome (ACLS) is an autosomal recessive syndrome with corpus callosum agenesis, occasional anencephaly and/or Dandy–Walker malformation, hypertelorism, post-axial polydactyly of the hands and pre-axial polydactyly of the feet[1] caused due to mutations in *KIF7* gene.[2] ACLS is extremely rare in Indian population.[3,4] Literature available regarding anaesthesia exposure in patients with these syndromes is scarce.[5] Anaesthetising a patient with an uncommon genetic disorder with multiple anomalies can prove to be a real challenge. We report a child born with the features of ACLS, who underwent multiple uneventful anaesthesia exposures.

CASE REPORT

Our patient was born to a 30-year-old primigravida at 36 weeks of gestation. She weighed 2.574 kg with an Apgar score of 6. The patient's mother booked into our hospital at the 35th week of pregnancy for safe confinement, when the in-house antenatal scan showed polyhydramnios with evidence of hydrocephalus and vermian hypoplasia. Elective caesarean section was planned, with the family deciding to continue active resuscitation of the baby. The baby had a weak cry and respiratory distress at birth requiring endotracheal intubation with a video laryngoscope (C-MAC® KarlStorz 8401BXC Mittelstrasse8 D-78532 Tuttlingen Germany). The child had bilateral cleft lip and palate, hypertelorism, short long bones, bilateral congenital talipes equinovarus (CTEV) and lower limb hallux duplex [Figure 1]. Cranial ultrasonography showed evidence of hydrocephalus and vermian hypoplasia. Magnetic resonance imaging (MRI) of the brain [Figure 2] showed partial callosal agenesis, pachygyria, ventriculomegaly, agenesis of cerebellar vermis and Dandy–Walker malformation. Based on the clinical features and cranial ultrasound and MRI findings, a diagnosis of ACLS was made. Echocardiogram revealed a patent ductus arteriosus (PDA) and patent foramen ovale (PFO). Genetic assessment revealed a large duplication in the *KIF7* gene which was consistent with genetic diagnosis of ACLS. The infant received mechanical ventilation for 2 days and the trachea was extubated uneventfully on the 3rd day. In view of persistent respiratory distress and increased oral secretions, the patient underwent fibreoptic laryngobronchoscopy in the Neonatal Intensive Care Unit (NICU) and was found to have severe laryngomalacia and laryngeal oedema probably secondary to gastro-oesophageal reflux (GER). The baby was treated with anti-reflux therapy and short course of nebulised steroids for laryngeal oedema. A more definitive treatment of laryngoplasty was deferred due to lack of parental consent. In view of recurrent GER, gastrostomy was planned under general anaesthesia (GA) at the age of 3 months. Tenotomy was done for CTEV on the 56th day of life under spinal anaesthesia. Eutectic mixture of lidocaine and prilocaine (0.5 g) was applied over L4-L5 anatomical landmark to provide topical anaesthesia. With the child breathing spontaneously on O_2-air mixture (50:50), adequate lateral positioning was maintained without neck flexion to avoid airway obstruction. Spinal anaesthesia was administered with a ½-inch 26-gauge Becton Dickinson Precisionglide™ needle by midline approach with 0.3 ml of 0.5% heavy bupivacaine. The child was haemodynamically stable throughout the procedure and tolerated the procedure well. At 3 months of age, in view of recurrent GER, gastrostomy under a non-opioid, non-relaxant, sevoflurane-based GA (maintaining adequate MAC) was conducted. A 4-mm uncuffed portex tube was used to secure the airway using video laryngoscopy. Analgesia was provided with paracetamol 15 mg/kg and 0.25% bupivacaine (up to 2 mg/kg) local infiltration. The child had uneventful anaesthesia, was extubated on the table and was observed in

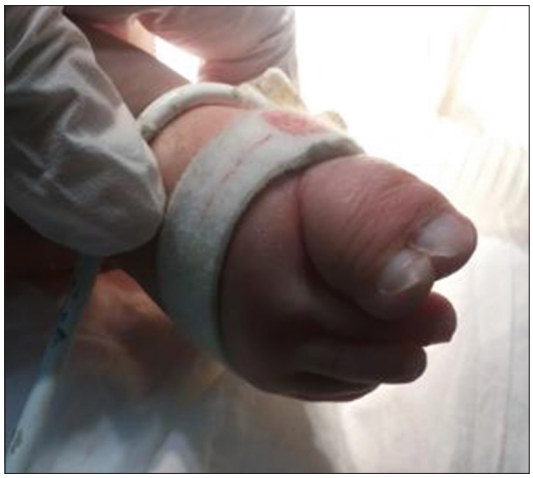

Figure 1: Hallux duplex

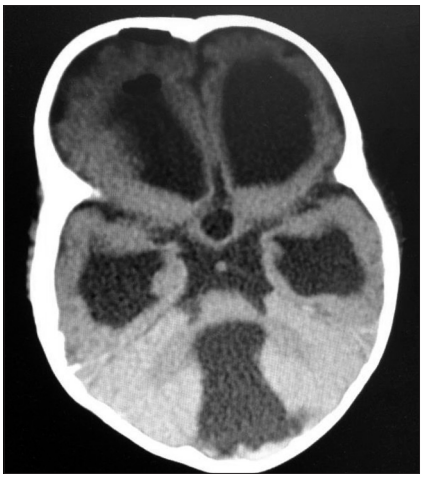

Figure 2: Radiographic evidence of the absence of corpus callosum

the NICU post-operatively. She underwent Nissen fundoplication at 4 months of age, cleft lip repair at 6 months age and cleft palate repair at 18 months of age using the same technique. During the last two anaesthetic exposures, the child was also administered fentanyl (1 mcg/kg) for analgesia. On all occasions, she tolerated the general anaesthetic exposures well, was extubated on the table and had an uneventful recovery. In the Paediatric Intensive Care Unit, the baby was started on high flow oxygen by nasal cannula therapy and shifted out of intensive care subsequently. Chest physiotherapy and saline nebulisations were included in her treatment schedule. Serial head scans and head circumference measurements were done to monitor hydrocephalus. At present, the child is referred to a neurodevelopment specialist, for neurodevelopment assessment and occupational therapist for infant stimulation program.

DISCUSSION

ACLS, also known by its synonyms: Schinzel ACLS and Hallux duplication, post-axial polydactyly and absence of corpus callosum, is a rare genetic disorder that is apparent at birth.[3] We found only one reference in literature for a child with ACLS being anaesthetised.[5] Facial anomalies such as micrognathia, hypertelorism, cleft lip and palate seen in these infants could lead to a 'difficult airway' scenario. Malformed or hypoplastic lungs with trachea or bronchi or both being stenotic or rarely dilated are frequent findings in these children. These features combined with abnormalities of respiratory control due to changes in the brainstem and cerebellum[4] would have been responsible for respiratory distress at birth requiring a video laryngoscope-guided endotracheal intubation. Fibreoptic laryngoscopy done 2 days after birth revealed laryngomalacia and laryngeal oedema attributed to gastro-oesophageal reflux prompting treatment with anti-reflux therapy and nebulised steroids. These patients are known to have congenital heart defects in the form of a large ventricular septal defect combined with atrial septal defect. Echocardiogram in our patient revealed PDA and PFO. These patients may have club feet, short extremities, abnormal knee joints, polydactyly, post-axial in the hands and pre-axial in the feet, with a typically duplicated big toe. Our patient had CTEV and bilateral lower limb hallux duplex. Hydrolethalus syndrome, Joubert's syndrome, oral-facial-digital syndrome, etc., were differential diagnoses in this patient. These patients are extremely sensitive to the respiratory depressant effects of general anaesthetic agents. Anaesthesia using inhalational induction, controlled ventilation, avoidance of opioids and close post-operative monitoring is recommended for these infants. Regional anaesthesia is also an available option.[6,7] The child underwent tenotomy under spinal anaesthesia at 56th day of life uneventfully, in an attempt to avoid the risk of administration of GA.[8] The child underwent multiple uneventful general anaesthetics at the age of 3, 4 and 6 and 18 months for gastrostomy, Nissen fundoplication, cleft lip and palate surgeries, respectively, with no requirement for post-operative mechanical ventilation. For final diagnosis, next-generation sequencing (NGS)[9] revealed a large duplication in the *KIF7* gene confirming the diagnosis of ACLS.

CONCLUSION

Sevoflurane-based anaesthesia with judicious use of opioids and spinal anaesthesia performed with due care formed the basis of an uneventful anaesthesia management, thereby avoiding anaesthesia-related complications in an a child with cardiovascular and nervous system defects.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given her consent for images and other clinical information to be reported in the journal. The guardian understands that names and initials will not be published and due efforts will be made to conceal patient identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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