

Anesthetic care of Roberts syndrome: A Case report

J Khandrani, A Rajput, A Khan, M Nagrale

Citation

J Khandrani, A Rajput, A Khan, M Nagrale. *Anesthetic care of Roberts syndrome: A Case report*. The Internet Journal of Anesthesiology. 2009 Volume 24 Number 2.

Abstract

Roberts Syndrome is a rare genetic disorder, characterized by multiple systemic involvement. In this report we describe the presentation and management of a case of Roberts syndrome. We report this case of a 4 year old boy who presented with tetraphocomelia, operated cleft lip & palate, mild mental and growth retardation posted for below knee amputation. We administered a subarachnoid block with light general anesthesia using laryngeal mask airway without any complication.

INTRODUCTION

Robert-SC syndrome is a rare inherited disorder characterized by nearly symmetrical reductive malformations of the limbs resembling phocomelia. Due to phocomelia-like limb defects similar to those seen in thalidomide embryopathy, the syndrome is also known as the pseudothalidomide syndrome. It is an autosomal recessive disorder with great variability of expression within families first described by Roberts(1)

<http://www.ijps.org/article.asp?issn=0970-0358;year=2008;volume=41;issue=2;spage=222;epage=225;aulast=Murthy-ref1> Later, Herrmann et al (2) described a very similar entity called pseudothalidomide or SC Syndrome. Zergollern and Hitrec (3)

<http://www.ijps.org/article.asp?issn=0970-0358;year=2008;volume=41;issue=2;spage=222;epage=225;aulast=Murthy-ref3> concluded that Roberts Syndrome and SC phocomelia are considered the same entity, and therefore, termed it as the Roberts-SC Phocomelia syndrome. These patients have flexion contractures of various joints, micrognathia, scanty hair, delayed growth, microcephaly with or without mental retardation, haemangiomas especially in the head and neck region, corneal opacity, ear anomalies, and may have cleft lip and palate. The typical hand and foot anomalies include an absence of the thumb, shortening of the first metacarpal bone, hypoplasia of the first digit, fusion of the fourth and fifth metacarpals, clinodactyly of the second and fifth digits, and hypoplasia of the middle phalanges. The upper limb is often more severely affected than the lower limb. Other less common features are cryptorchidism, an enlarged phallus (compare to the rest of body), oligohydramnions, renal anomalies (polycystic or dysplastic), heart anomalies (ASD,

PDA), and gastrointestinal abnormalities.

CASE REPORT

A 4 year old boy, 10kg, fourth child of 2nd degree consanguineous marriage of parents was admitted with chief complaints of defect in all four limbs, palate & upper lip since birth and delay in speech .

On examination all four limbs were like buds. The upper limbs were present up to 2 cm below the cubital fossa and ended into a thumb like structure on the radial side. The lower limb ended into the great toe like structure on the tibial side. He had a repaired left sided unilateral cleft lip, cleft palate and low set ears. Head, eyes, spine and genitalia were normal. His systemic examination including cardiovascular and respiratory system were normal. On airway assessment he had large head with adequate mouth opening and large tongue with incomplete cleft palate.

Haematological (Hb, TLC, DLC, PS), biochemical (kidney functions , liver functions) investigations, USG of abdomen, 2- D echo-cardiographic evaluation of heart were in normal limits.

As the child had haememelia i.e. congenitally both tibia were malformed so child was posted for below knee amputation. The child was nil by mouth before operation. He was shifted in OT in a warm blanket. With pulse oximeter and ECG monitoring the patient was induced with incremental doses of Halothane applied by face mask. Monitors including cardioscope, temperature and capnography were attached. It was difficult to insert an IV line as all four limbs were malformed. So an IV line was secured with a 22G vasofix in the external jugular vein. Inj.

glycopylorrate 40mcg and Inj. midazolam 0.5mg were given as premedication. Inj. Ketamine 10mg IV was given. The patient was mask ventilated with an oral airway and a LMA of size 1.5 was inserted. Under all aseptic precaution the patient was given a subarchnoid block using 1cc of 0.5% bupivacaine. After 8 mins, the level of subarchnoid block was checked using pin prick. No tachycardia was observed with pin prick at T10 level. Surgery started and lasted for 60mins. Blood loss during procedure was within permissible limit. There were no significant intra-operative events and vitals parameters were maintained. Halothane and Nitrous oxide were tapered at the end of surgery. LMA was removed with adequate suctioning and cuff deflation. When good muscle tone and response to verbal command achieved. The child was shifted to recovery room for observation.

Figure 1

Figure1: Preoperative photograph of Roberts Syndrome



Figure 2

Figure2: Intra-operative picture of Roberts Syndrome



DISCUSSION

Roberts SC syndrome is a rare syndrome. It was initially described by Roberts in 1919 (1) as Roberts Syndrome, and reviewed by Appelti (4). Herrmann described in 1974, cases of a very similar entity called pseudothalidomide or SC syndrome (OMIM: 269000) named after the initials of the surnames of the two families (SC) in whom it was originally described (5).

Most patients born at term with less than 37 cm of birth length and severe facial and limb defects have been still born or have died early in childhood. Patients born with more than 37 cm of birth length and less severe defects have better prognosis. However, survival beyond infancy is infrequent in patients affected by this syndrome (6).

Roberts syndrome is a rare genetic disorder, characterized by multiple malformations, in particular symmetrical limb reduction, craniofacial anomalies, such as bilateral cleft lip and palate, nose and ear anomalies, and severe mental and physical retardation. This patient are challenge for anesthesiologist as it is associated with difficult airway, heart defects (ASD and PDA), renal anomalies (polycystic or dysplastic kidneys), limb defects and gastrointestinal abnormalities (obstructions). So whenever such patient is encountered detail evaluation of every system is needed.

Luckily our patient when investigated did not reveal any systemic involvement including echo-cardiography was within normal limit. With all limbs being malformed it is always difficult to secure I.V. line. Also with various cranio facial deformities including macroglossia, partial cleft palate and retrognathic mandible it is difficult to mask ventilate and

intubate. So advanced airway aids should be in hand before anesthetizing such patient.

In our case mask ventilation was difficult due to large tongue but it was overcome by putting oral airway during mask ventilation followed by LMA. As below knee amputation is major surgery to reduce intra-op anaesthetic requirement and analgesia, subarachnoid block was given which provided stable haemodynamic profile throughout intra-operative period.

In this way, a difficult case of rare congenital anomaly was managed with light general anesthesia and subarachnoid block which gave stable hemodynamic vitals.

References

1. Roberts JB. A child with double cleft of lip and palate, protrusion of the intermaxillary portion of the upper jaw and imperfect development of the bones of the four extremities. *Ann. Surg.*, 1919, 70: 252
2. Herrmann J., Optiz J M: The Sc phocomelia and the Roberts Syndrome: nostalgic aspect. *Eur. J Paediatr.* 125:117-134, 1977
3. Zergollern L, Hitrec V. Four siblings with Roberts syndrome. *Clin Genet* 1982;21:1-6.
4. Appelt H, Gerken H, Lenz W. Tetrachokomelie mit Lippen-Kiefer-Gaumenspalte und Clitorishypertrophie - Ein Syndrom. *Paediatr. Paedol.*, 1966, 2: 119
5. Herrmann J, et al. A familial dysmorphogenetic syndrome of limb deformities, characteristic facial appearance and associated anomalies: The pseudothalidomide or SC-syndrome. *Birth Defects*, 1969, 5: 81
6. Murthy J, Dewan M, Hussain A. Roberts-SC syndrome, a rare syndrome and cleft palate repair. *Indian J Plast Surg* 2008;41:222-5

Author Information

Jitesh Khandrani

Assistant Professor, Department of Anesthesiology Datta Meghe Institute of Medical Science, Sawangi(Meghe), Wardha, India

Anant Rajput

Assistant Professor, Department of Anesthesiology Datta Meghe Institute of Medical Science, Sawangi(Meghe), Wardha, India

A. Khan

Associate Professor, Department of Anesthesiology Datta Meghe Institute of Medical Science, Sawangi(Meghe), Wardha, India

Manda Nagrale

Professor & Head, Department of Anesthesiology Datta Meghe Institute of Medical Science, Sawangi(Meghe), Wardha, India