ORIGINAL RESEARCH PAPER
Volume - 12 | Issue - 05 | May - 2023 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

ANAESTHETIC MANAGEMENT OF A CHILD WITH ROBINOW SYNDROME

Anaesthesiology

Dr. Prem Chandar. L. K Postgraduate in Department of Anaesthesiology and Pain medicine, Saveetha medical college and hospital, Chennai.

Dr. Navaneetha Krishnan Professor, Department of Anaesthesiology and Pain medicine, Saveetha medical college and hospital, Chennai.

Dr. Rohit Kamal Postgraduate in Department of Anaesthesiology and Pain medicine, Saveetha medical college and hospital, Chennai.

Dr. Lakshmi Ramakrishnan Head of the department of anaesthesia and pain medicine, Saveetha medical college and hospital.

ABSTRACT

Robinow syndrome (Robinow-silverman-smith syndrome or fetal face syndrome) is a rare disease characterised by anomalies in face, head, reproductive organs and spine segmentation. Anaesthetic challenges may include difficult airway due to structural anomalies of face and neck, congenital cardiac or renal abnormalities and their preoperative optimisation. We report the anaesthetic management of a patient with Robinow syndrome presenting for cleft palate surgery. The child presented with, web neck, partially fused ribs, fetal facies (small face and widely spaced eyes), prominent forehead, with associated patent ductus arteriosus. Robinow syndrome usually presents with features of fetal face appearance, genital hypoplasia and gingival hypoplasia and shortening of limbs. Around 15% population will have congenital heart defects. Other common features include brachymelia, hemivertebrae, dysmorphic face, genital hypoplasia, micropenis, clinodactyly, camptodactyly, hypoplastic fingers and short stature. Cardiac defects and hepatosplenomegaly can be observed. Hence in all patients difficult airway should be anticipated and difficult airway cart should be ready. Associated congenital anomalies should be ruled out and optimised accordingly.

KEYWORDS

Robinow syndrome, cleft palate, difficult laryngoscopy, intubation, congenital, consanguineous

INTRODUCTION

Robinow syndrome is a rare inherited disorder that generally affects the development of bones and other parts of the body. There are two forms of Robinow syndrome: Autosomal Dominant, which is milder and Autosomal recessive which is more severe. Main symptoms of this syndrome are mesomelic limb shortening, abnormal facial morphogenesis, hemivertebrae, genital hypoplasia, hypoplastic fingers. Robinow syndrome patients present with surgeries for skeletal, cardiac anomalies or cleft surgeries. We describe the anaesthetic management of a case of Robinow syndrome presented with cleft palate.

CASE REPORT

The patient was a 1 year and 4 months old female (weight 9 kg) who was a diagnosed case of isolated cleft palate defect and a known case of Robinow syndrome. Mother complained of child having frequent nasal regurgitation, clear nasal discharge and snoring during sleep.

Antenatal history revealed
- Delayed perception of fetal movements
- Polyhydramnios

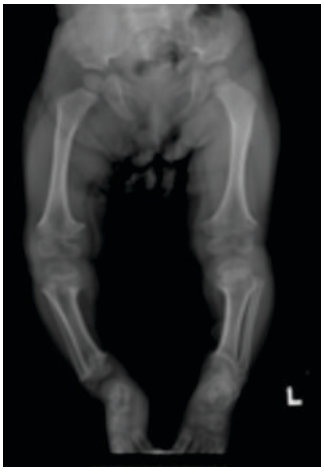

Figure 1 Radiograph of lower limbs showing mesomelic limb shortening.

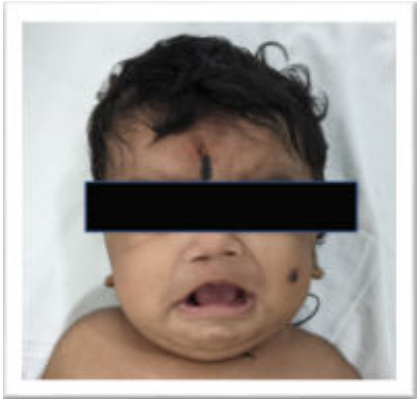

Figure 2 Front view of the baby showing the features of hypertelorism and short neck (*Picture was taken and published here after obtaining informed and written consent form the parent*)

The child was born out of a III degree consanguineous marriage. Preoperative assessment including complete blood count, renal function tests revealed normal laboratory values. Additionally Bleeding and clotting time was done to assess the clotting status. Airway assessment revealed decreased range of movements of neck, thick and short neck, hypertelorism. COPUR score was 8 indicating mild difficulty in intubation.

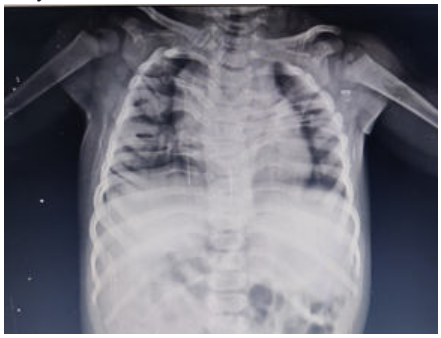

Figure 3 Chest radiograph showing crowding ribs

Head to toe examination revealed fetal facies with prominent forehead, hypertelorism, crowding bifid hypoplastic ribs with scoliosis, short neck, bilateral clinodactyly, bilateral hypotonia, mesomelic limb shortening. The child had adequate cry.

Chest radiograph revealed pulmonary hypoplasia of left upper lobe. Echocardiography revealed Patent ductus arteriosus with left to right shunt. Patient was shifted inside operating room. Standard monitoring equipment were applied which includes Noninvasive Blood pressure, pulse oximeter and ECG. Difficult airway cart was made ready. Anaesthesia was induced with sevoflurane and oxygen using step-up technique (1-8%) via face mask and mask ventilation was possible. IV line secured and IV fentanyl 18 mcg given. A check laryngoscopy was done to assess the Cormack-Lehane grade using Mcintosh size 2 blade. But the CL grade was 3. Hence miller size 2 blade was used, and the arytenoids were visualized. Before laryngoscopy a cloth roll was placed in the interscapular region to achieve the sniffing position since the head to body ratio was large compared to adults. Hence the child was paralyzed using atracurium and laryngoscopy was done but the passage of endotracheal tube was difficult. Hence, we adopted Para-glossal approach to intubation using miller blade size 2 in which we were able to pass a bougie. Using bougie, trachea was intubated with 3.5 mm cuffed tube. Anaesthesia was maintained with Sevoflurane, Oxygen and Air. The surgery duration was 1.5 hours and post operatively extubation was uneventful.

DISCUSSION:

Robinow syndrome is a rare inherited disease that may present in autosomal recessive or dominant forms. Of these two overlapping phenotypes, the recessive form has been described as having more severe skeletal deformities. The gene responsible for the autosomal recessive form is located at the long arm of chromosome 9, and the **ROR2 tyrosine kinase** is thought to be the enzyme involved in the pathogenesis. Robinow syndrome usually occurs in areas featuring a high level of consanguinity. Features include mesomelic limb shortening, hypertelorism, midfacial hypoplasia, short, upturned nose, broad forehead and nasal root, 'tented' upper lips, occurrence of cleft lips and palate, pseudo-exophthalmos from lower eyelid deficiency, spinal deformities, hypoplastic genitalia, kidney disease and congenital heart defects. Airway difficulty must always be anticipated in any case of Robinow syndrome, and a difficult airway trolley should be readily available. Skeletal abnormalities may increase the risk of failed neuraxial blockade in Robinow patients. Severe kyphoscoliosis may affect chest wall mechanics and pulmonary gas exchange, increasing perioperative risk. Renal abnormalities such as hydronephrosis and cystic kidney dysplasia may present. These patients are prone to develop Urinary tract infections. Congenital heart defect is the major cause of death during the initial years of life in these patients. Pulmonary stenosis or atresia are the most common defects, followed by septal defects, coarctation of the aorta, tricuspid atresia, and tetralogy of Fallot. These heart defects increase the perioperative risk.

The following preoperative workup is proposed for patients with Robinow syndrome:

- ECHO to evaluate cardiac defects and vessel stenoses.
- Laboratory investigations to include blood urea, nitrogen, creatinine, liver function tests, full blood count and coagulation status, and urinalysis for possible UTI.

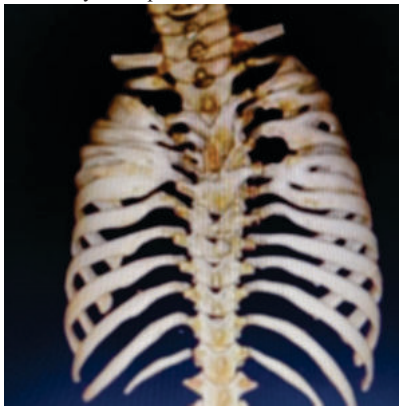

Figure 4 Three-dimensional reconstruction of thorax showing severe deformity of the ribs

CONCLUSION:

Hence in Robinow syndrome, the challenge to an anaesthetist doesn't end only with difficult airway. A comprehensive preoperative evaluation consisting of airway assessment, cardiac evaluation, imaging for chest and spine and renal function tests are mandatory. Operating theatre should be fully equipped to face all sorts of difficulty regarding the airway.

REFERENCES:

1. Frazier MS, Drzymkowski J. Developmental, congenital, and childhooddiseases and disorders. In Essentials of Human Diseases and Conditions. 5th ed., St. Louis: Elsevier Saunders, 2013:38-123.
2. Patton MA, Afzal AR. Robinow syndrome. J Med Genet 2002;39(5):305-310.
3. DeMauro SB, Wei JL, Lin RJ. Perspectives on neonatal and infant tracheostomy. Semin Fetal Neonatal Med 2016;21(4):285-291.
4. Macdonald I, Dearlove OR. Anaesthesia and Robinow syndrome. Anaesthesia 1995;50(12):1097.