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Robinow Syndrome : an anesthetic challenge and review of literature

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Abstract : Robinow Syndrome is characterized by the presence of mesomelic limb shortening, midfacial hypoplasia, hemivertebrae and genital hypoplasia. Multi-organ involvement including cardiac, renal, vertebral dysfunctions have been described.We here report successful anaesthetic management of 1year old boy posted for ophthalmic procedure.

Key words : Robinow syndrome ; pediatric anesthesia ; mid facial hypoplasia ; facial dysmophism ; ophthalmic surgery.

Introduction

Robinow Syndrome or Fetal Face Syndrome characterized by the presence of mesomelic limb shortening, midfacial hypoplasia hemivertebrae, and genital hypoplasia was first defined by Robinow Meinhard in 1969 (1, 2). Multi-organ involvement including cardiac, renal, vertebral dysfunctions, has also been described.

We present the case of a 1-year-old male child, a diagnosed case of Robinow Syndrome who was scheduled for an ophthalmic examination under general anesthesia and review the perioperative anesthetic concerns.

Case report

A one-year-old male child, weighing 7.5kg, born out of non-consanguineous marriage, was scheduled for ophthalmic examination under general anesthesia. He was a diagnosed case of Robinow Syndrome and presented with the classical limb shortening, hypertelorism, midfacial hypoplasia along with retrognathia, micrognathia, long philtrum, upturned nose, frontal bossing and large anterior fontanelle (Fig. 1A & B). His preoperative laboratory investigations and echocardiography were within normal limits.

After obtaining informed written consent, anesthesia was induced with 8% sevoflurane in 100 % oxygen while maintaining spontaneous respiration. A two-hand technique was used to achieve an adequate seal due to dysmorphic facial features. After achieving adequate of anesthesia, a 24G intravenous cannula was cannulated followed by Inj. Fentanyl 1µg/kg. An AMBU-LMA, size 1.0 was successfully inserted, and adequate depth of anesthesia was maintained with pressure support ventilation and Sevoflurane (Fig. 1C). The post-operative period was un-eventful. After completion, the AMBU LMA was successfully removed and the postoperative recovery was unremarkable.

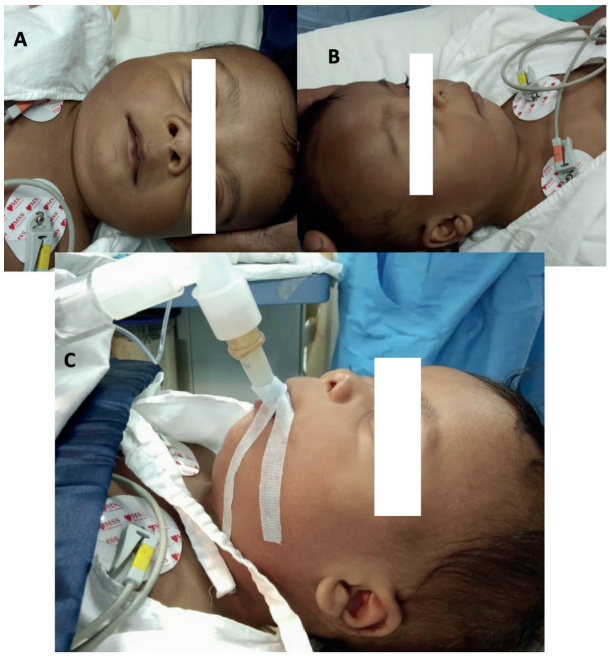

Figure 1. — A-Showing mid facial hypoplasia. B- Lateral view of face showing retrognathia and micrognathia. C-Showing successful LMA placement.

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Discussion

Robinow syndrome is a rare genetically inherited disorder. The incidence of Robinow syndrome is about 1:500,000 but prevalence is slightly lower as 5-10% of children die in infancy. The male to female ratio of Robinow syndrome is 1:1 (18). The syndrome is described in two forms, an autosomal recessive type with distinctive shortening of limbs, short stature, facial dysmorphism, and a milder autosomal dominant type with less severe abnormalities. The mutation in the ROR2 gene is responsible for the autosomal recessive type. Afzal et al. mapped the gene encoding receptor orphan receptor tyrosine kinase 2 (ROR2) to chromosome 9q22 (3, 4). Mutations in WNT5A, locus 3p14.3 (OMIM 164975) are responsible for the autosomal dominant (AD) form of RS. In these patients, oral manifestations are more prominent, hemivertebrae and scoliosis rarely occur and facial abnormalities tend to be milder.

Airway

Robinow Syndrome presents with characteristic facial dysmorphism that includes midfacial hypoplasia, hypertelorism, upturned nose, long philtrum, pseudo-exophthalmos due to lower eyelid deficiency (1,2). There is gingival hypertrophy with dental crowding. Choanal atresia (5), with cleft- lip, and palate (6) may also be observed. These features can lead to upper airway obstruction and difficulty in securing the airway (7). In our case, difficulty in the bag and mask ventilation were encountered, although the insertion of AMBU LMA was uneventful. A difficult airway cart should always be made available. Successful intubations have been reported by MacDonald et al and Lirk et al. (8, 9). However, a case report published by Weksler et al described difficult laryngoscopy and unsuccessful tracheal intubation in a case of Robinow Syndrome, nevertheless, they were able to use LMA as a rescue device to secure airway (10). Cassinello Ogea C et al also reported the successful use of LMA in a case of Robinow Syndrome with difficult airway and grade IV Cormack-Lehane (11).

Skeletal abnormalities

These patients may present with hemivertebrae leading to kyphoscoliosis, fused, or missing ribs leading to chest deformity (6). This may affect respiratory function and pulmonary gas exchange (9). Vertebral anomalies may prove a hindrance to regional anesthesia, and therefore should be evaluated beforehand.

MacDonald and Dearlove reported the use of a single-shot caudal block in a patient posted for psoas hitch procedure successfully. Except for increased sacral dimpling, the spine and pelvis were otherwise normal (8).

Cardiac Anomalies

A major concern for the anesthesiologists is the presence of congenital heart disease.

Patton reported pulmonary stenosis or atresia as the most common defect, followed by septal defects, coarctation of the aorta, tricuspid atresia, and tetralogy of Fallot (6, 12). The patient should be screened to rule out congenital heart disease and preoperative echocardiography should be done in patients with definite symptoms.

Renal and Genitourinary anomalies

Genital hypoplasia in terms of the hypoplastic clitoris and labia minora/Majora in females or a small penis with a normal scrotum and testis in males have been observed (13). Obstructive uropathy, renal cysts, hydronephrosis are other anomalies

Table I

Various organ system involvement and anesthetic implications in a patient with Robinow Syndrome

Organ involvement	Characteristic finding	Anesthetic implication
Airway	Midfacial Hypoplasia, retrognathia, micrognathia, choanal atresia, cleft lip and cleft palate, gingival hyperpalsia	Difficult airway, bag mask ventilation and intubation
Cardiovascular	Pulmonary stenosis, septal defects, coarctation of aorta, tricuspid atresia, Tetralogy of Fallots	Anesthetic implication as per cardiac lesion
Vertebral and Skeletal abnormalities	Mesomelic limb shortening, hemivertebrae, kyphoscoliosis, fused ribs	Difficulty in positioning, difficulty in giving regional block, respiratory complication
Genito-urinary	Genital hypoplasia, obstructive uropathy, renal cysts, hydronephrosis	Renal failure, drug dose modification
Eye	Pseudo-exophthalmos	Proper protection is needed
Central nervous system	Mental retardation, developmental delay	Judicious use of anaesthetic drugs
Liver	Association with Criggler-Najjar Syndrome	Proper evaluation of liver function is needed

(14). A preliminary renal function should be done to look for any derangement and further evaluation should include ultrasonography.

Central Nervous System

Intelligence is usually normal but mental retardation and developmental delay have been noted in up to 20 % of patients with Robinow syndrome (6).

Endocrinology

In male patients, low testosterone levels and impaired testosterone response to human chorionic gonadotropin stimulation are seen ; whereas in female patients, the hypothalamic-pituitary axis or the hormonal response of the ovaries seems to be impaired (15).

Hepatic abnormalities

An association with Crigler-Najjar has been reported in the literature, although not proven. A baseline liver function should be done to rule out any abnormality (16, 17).

Conclusion

A patient with Robinow Syndrome may prove as a challenge to the anesthesiologists due to difficult airway and the high incidence of congenital heart disease. A thorough physical examination and history along with necessary investigations preoperatively with proper preparation for difficult airway can help improve the outcome in these patients.

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