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A Rare Entity: Case Report on Anaesthetic Management in Robinow Syndrome

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Abstract: ***Robinow syndrome (RS) is an infrequent genetic disorder with an incidence of 1: 500, 000 which clinically presents with short stature, cardiac, genitourinary, and skeletal deformities, as in this case who had butterfly vertebrae at lower thoracic and lumbosacral region - making caudal block extremely difficult.***

Keywords: Robinow syndrome, genetic disorder, short stature, caudal block, paediatric anaesthesia

1. Case Description

A five - year - old male child presented with complaints of growth retardation and no appreciable increase in the penile size since birth. Born from non - consanguineous marriage After thorough preoperative evaluation, optimisation and obtaining written informed parental consent, he was posted for de - gloving of the penis with dissection of corpora under anaesthesia.

In the preoperative examination of the patient, facial appearance peculiar to RS, Mallampati III, reduced anteroposterior diameter of the chest. Laboratory values, electrocardiography (ECG) and 2D - Echocardiography (ECHO) were normal. An anteroposterior roentgenogram of thoracic and lumbosacral spine showed butterfly and hemivertebrae at multiple levels. The patient was considered to be ASA II. On the day of surgery after ensuring fasting of 6 hours patient was taken to the operating theatre, and monitoring was done with ECG, non - invasive blood pressure, peripheral oxygen saturation (SpO_2) and body temperature. Heart rate was 100 beats per minute, blood pressure was 88/56mmHg, and SpO_2 was 99% at room air. The patient was pre - oxygenated with 100% oxygen with an anatomical face mask. After induction of anaesthesia with 1 - 8% Sevoflurane, a 24G peripheral venous catheter was placed in the dorsum of the right hand and intravenous fluids were started. The patient was pre - medicated with 1 mic/kg of fentanyl, and the airway was secured with a supraglottic airway device. For anaesthesia maintenance, 2% Sevoflurane with air in a 50: 50 ratio was used. Ultrasonography (USG) guided caudal block was given. No neuromuscular blocks were used. The supraglottic airway device was removed at the end of surgery while the patient was monitored in the anaesthesia care unit. He was discharged the next day and followed up at regular intervals.

2. Discussion

Genetically RS can be autosomal dominant, autosomal recessive or heterogenous. The most important manifestations are due to mutation in the autosomal recessive gene ROR2, which is located on chromosome 9q22. Its frequency is 1: 500, 000, and it is equally observed in both sexes. In these cases, surgical treatment may be needed for vertebral, genitourinary orofacial and dental anomalies. Mental and motor development in these cases is generally normal as was in our case.

3. Summary

Robinow syndrome RS is a rare genetic disorder with an incidence of 1: 500, 000, characterised by short stature, cardiac, genitourinary, and skeletal deformities. This case study involved a five - year - old male with growth retardation and underdeveloped penile size. Born from a non - consanguineous marriage, the child presented with characteristic facial features of RS, Mallampati III classification, and reduced anteroposterior chest diameter. Despite normal laboratory values and cardiac assessments, imaging revealed multiple butterfly and hemivertebrae. Classified as ASA II, the patient underwent penile de - gloving and dissection under general anaesthesia.

Preoperative and intraoperative management included ECG monitoring, pre - oxygenation, induction with Sevoflurane, and a USG - guided caudal block. The surgery proceeded without complications, and the patient was discharged the next day. RS, caused by mutations in the ROR2 gene on chromosome 9q22, presents unique challenges in anaesthesia due to its rare occurrence and associated anomalies.

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