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Anaesthetic implications of Rubinstein–Taybi syndrome

Rubinstein Taybi Syndrome (RTS) (broad thumb-hallux syndrome) is a rare (1/250,000–300,000) autosomal dominant genetic disorder. It is characterised by mental retardation, physical abnormalities, including broad thumbs, big and broad toes, short stature and craniofacial abnormalities [1]. To date there is limited literature on anaesthetic management of RTS. We describe the anaesthetic management of a child of RTS for left eye therapeutic syringing and probing. Written Informed consent to publish this case report was obtained from the child's guardians.

A 5 year old child (15 kg, 94 cm) of RTS presented to Ophthalmology department, with left eye discharge was planned for nasolacrimal duct syringing and probing. Child was born of non-consanguineous marriage, had delayed crying and delayed milestones. On gross examination he had short stature, high arched eye brows, frontal bossing and micrognathia. Oral examination revealed mal aligned teeth, high arched palate and grade II-III tonsils. Skeletal examination revealed broad thumbs and toes, hand and foot abnormalities, pectus carinatum, scoliosis and flat foot (Fig. 1). On cardiac evaluation, he was found to have congenital heart disease (PDA 1.1 mm left to right shunt with PFO 1 mm ASD).

Recent cardiac evaluation revealed small PDA with no ASD with normal biventricular function. Rest investigations were unremarkable.

Routine American Society of Anaesthesiologist (ASA) monitors were applied. His vital parameters were: Heart rate 120/min, saturation 99% on room air. Patient was induced with incremental doses of sevoflurane with 50% air and oxygen at 6 lt/min flows and Intravenous line was secured on left hand with 24 G cannula after achieving adequate depth of anaesthesia. Airway was secured with 1.5# AMBU Laryngeal mask airway.

Anaesthesia was maintained with sevoflurane 2–3% and 50% oxygen in air with pressure support ventilation. Inj paracetamol 220 mg i/v was used for analgesia and hydration was maintained with 50 ml 1% in ringer lactate to avoid hypoglycaemia. Surgery lasted for 10 min and perioperative course was uneventful. Child was transferred to the post-anaesthesia care unit (PACU) for observation of hemodynamic and respiratory functions.

RTS results from sub microscopic 16p13.3 deletion of cyclic AMP response element-binding protein (CREBBP) gene [2,3]. Anaesthetic management of such patients is required for surgical correction of craniofacial, orthopaedic, orthodontic, ophthalmic or cardiac lesions.

There are several aspects of this condition that are important for anaesthesiologists. Multisystem involvement like airway, cardiac, musculoskeletal, respiratory and urogenital system poses challenge to anaesthesiologist for managing such cases. The airway management is challenging in such patients in view of craniofacial abnormalities, laryngotracheal abnormalities, obstructive sleep apnea, gastroesophageal reflux disease, increased risk of aspiration, mental retardation, and increased risk of arrhythmia with succinylcholine [4]. The abnormalities of head and face include microcephaly, antimongoloid faces, high

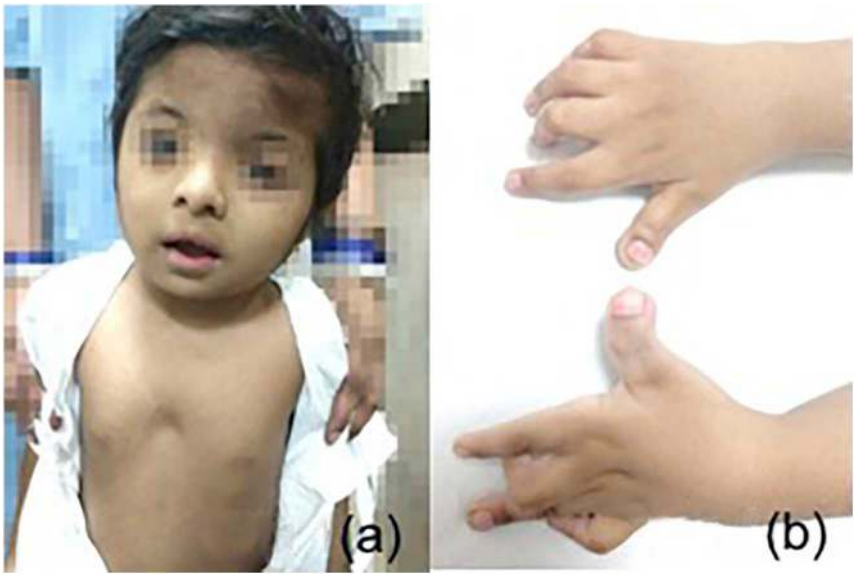

Fig. 1. (a) Child with frontal bossing, high arched eye brows, and micrognathia and pactus carinatum. (b) Broad thumb and fixed flexion finger abnormalities

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arched palate, widely spaced eyes, hypo plastic lower jaw with small mouth opening and dental abnormalities [4] which makes airway management challenging. Sedatives have to be avoided as premedication as child had history of snoring. Presence of patent ductus arteriosus necessitates vigilance while drug administration as avoiding air bubbles in intravenous line is necessary to prevent paradoxical air emboli [5]. Succinylcholine should be avoided in these patients because it is responsible for supraventricular tachycardia and early atrial and ventricular contractions [5].

To summarise, patients with RTS needs through preoperative evaluation for the multisystem involvement. Intense vigilance and monitoring in the perioperative period is needed for the successful management of a child with RTS.

Conflict of interest and source of funding

None to report.

Patient consent

Written consent was obtained from the patient guardians for publication of this report.

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