Case Report

A Comprehensive management of SATB2-Associated Syndrome (Glass Syndrome) for Vitrectomy: Careful Combat

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Abstract

SATB2-associated syndrome, also known as GLASS syndrome, is a recently identified condition characterized by developmental delay or intellectual disability, distinctive dysmorphic features, behavioural issues, absent or limited speech, and craniofacial abnormalities. This case report underscores the critical need for comprehensive preoperative evaluation, careful intraoperative monitoring, and strategic airway management planning in patients with Glass syndrome to ensure safe and effective anaesthetic care.

Keywords

dental defects, craniofacial abnormalities, family counselling, difficult airway

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Introduction

SATB2-associated syndrome (SAS), also referred to as Glass Syndrome, is a rare genetic disorder affecting multiple systems. It results from pathogenic variants in the SATB2 gene located on chromosome 2q33.[1] Although awareness of SAS has increased since its discovery, it remains underdiagnosed due to its rarity and diverse presentation.

The SATB2 gene can be altered through various mechanisms including point mutations, translocations, intragenic deletions, duplications, and contiguous deletions.[2] Its multisystem nature necessitates a thorough approach for assessment and treatment. Delays in neurocognitive development, craniofacial deformities, and dental problems are important characteristics.

Most common finding in this syndrome is Developmental delay / intellectual disability. Common eye findings are strabismus (18%) and refractive errors (8%).[1] Joint contractures, tibial bending, and scoliosis

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are examples of skeletal anomalies. Additional characteristics could include eating issues and growth restriction before and after birth. Additional symptoms like cardiovascular, genitourinary, and ectodermal abnormalities may also be seen in cases where the deletion includes SATB2 and neighbouring genes.

Case report

A 17-year-old female (body weight of 54 kg) with a history of SATB2-associated syndrome presented for a pars plana vitrectomy (PPV) procedure. Her medical history included sleep disturbances, praxis difficulties, minimal language skills, profound intellectual disability, and autism-like behaviour. The genetic analysis revealed a deletion on chromosome 2q32-q33, which is characteristic of this rare syndrome.

She was the only child of a couple, born at full term following an uneventful pregnancy. Dysmorphic features noted at birth prompted further investigations, which identified a chromosomal variation involving chromosome 2 as the initial diagnosis.

Her features included a long, thin face, a small chin, a drooping lower lip, a large forehead, downward and outward slanting palpebral fissures, and ears placed lower than usual, Figure1.

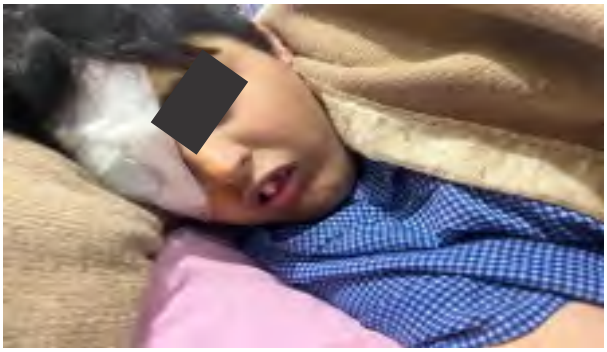

Figure 1 Facial features and craniofacial abnormalities

The patient had dental anomalies (irregularly sized or shaped teeth, supernumerary teeth, missing teeth) and a high-arched palate from the perspective of the airway. The patient's neurocognitive symptoms included a worldwide developmental delay, a decline in motor and communication abilities, angry and anxious episodes, sleep issues, disordered behaviour, increased hyperactivity and self-harming activities, Figure 2.

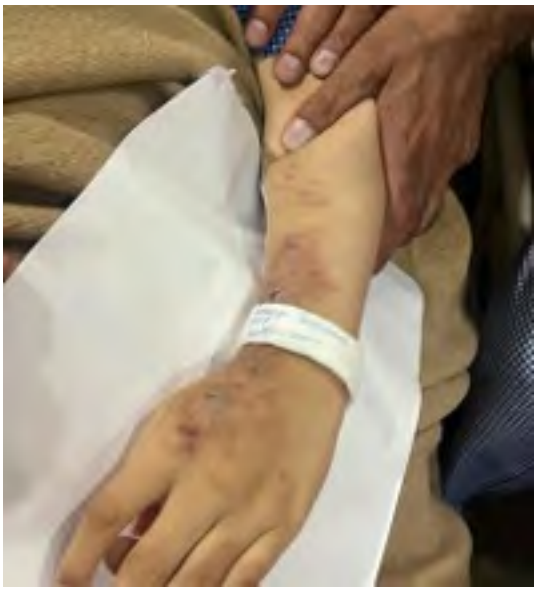

Figure 2 Self-mutilated injuries seen over forearm

Given these factors, significant anaesthetic concerns were identified, particularly related to airway management.

A thorough plan was formulated to address the possibility of a difficult airway. Use of C-MAC blade video laryngoscope was Plan A; the initial tracheal intubation plan. Secondary airway management plan (Plan B) was to employ a laryngeal mask (LM) either as a definitive airway or as a conduit for fibre-optic bronchoscopic (FOB) intubation.

In the pre operative room, a 20g intravenous (IV) cannula was successfully placed with parental assistance.

Preoperative sedation was achieved using IV midazolam (2 mg, given in incremental doses of 0.5 mg at intervals). After administering sedation, the patient was carefully transferred to the operating theatre, where standard monitors (pulse oximetry, non-invasive blood pressure, and electrocardiography) were attached. Baseline vitals were normal; heart rate 87 beats/minute, blood pressure 126/88 mm Hg and SpO2 100%. In anticipation of potential airway challenges, a fully equipped difficult airway cart was prepared, including advanced airway management tools.

Anaesthesia induction was initiated with intravenous fentanyl (110 g) and propofol (100 mg slow iv, titrated to loss of consciousness). After successful bag-mask ventilation a muscle relaxant was administered to facilitate tracheal intubation. Intubation was accomplished successfully using a C-MAC video laryngoscope, highlighting the significance of thorough planning and the application of cutting-edge airway management tools. Anaesthesia was maintained with O2(50%) and N20 (50%) mixture. Desflurane was the inhalational agent used and MAC was maintained between 1 to 1.2.

The patient was extubated following the reversal of muscle relaxation with neostigmine (0.05mg/kg) and glycopyrrolate (0.01 mg/kg) after the return of adequate spontaneous breathing. Post-extubation, patient was transferred to the recovery room, where close monitoring of vital signs and respiratory function was maintained to ensure a smooth recovery process.

Discussion

The diagnosis of GLASS syndrome is made more difficult by the fact that its clinical symptoms overlap with those of other hereditary diseases. Confirmation requires expert geneticists to sequence the exome. As an epigenetic regulator in neurodevelopment, the SATB2 gene, which is found on chromosome 2q, is essential for cognitive development and craniofacial patterning.3 Intellectual incapacity, dental crowding, hypodontia, delayed dentition, and diastema are common clinical features.

The patient's propensity for self-harming behaviour makes routine clinical procedures including intravenous (IV) cannulation difficult. Counselling, pre-operative preparation and assistance are necessary to manage such behaviours.

Multidisciplinary referrals to geneticists, paediatric neurologists, orthopaedic/dental surgeons, psychiatrists, physical and speech therapists are needed for comprehensive care and pre-operative stabilisation. Effective communication among team members prevents fragmentation of care.

A difficult airway trolley with video laryngoscopes (e.g., C-MAC), supraglottic devices, and a fibre-optic bronchoscope should be readily available. In many cases, direct laryngoscopy may be difficult due to distorted anatomy, making video laryngoscopy a preferred approach. In extremely challenging cases, fibre-optic intubation may be necessary to visualize the airway and secure it safely.

An early diagnosis based on genetic confirmation is essential for children with syndromic features. Perioperative problems can be decreased by using a customised anaesthesia strategy and being well-prepared for any airway issues. Multidisciplinary collaboration during pre-operative evaluation are essential to ensure patient safety and the best possible outcomes. In order to enhance the diagnosis, treatment, and quality of life for those with GLASS syndrome, this example emphasises the necessity of continued study and clinical awareness.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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