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Case Report

Anaesthesia Section

Anaesthesia in a Child With 9q13-q21 Deletion Syndrome: Tailored Perioperative Management in a Rare Genetic Disorder

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ABSTRACT

Rare genetic disorders may present unique challenges in the treatment of anaesthesia due to their variable clinical presentations and potential for unexpected complications, necessitating a highly customised approach to perioperative management. The 9q13-q21 microdeletion syndrome is an exceptionally rare chromosomal disorder with limited literature documentation. This case report describes the successful perioperative anaesthetic management of a 12-month-old male child with heterozygous microdeletion in chromosome 9q13-q21.33 who presented for orchidopexy. The patient exhibited developmental delay, profound hypotonia, and distinctive craniofacial dysmorphic features including microcephaly, midfacial flattening, hypertelorism, and blepharophimosis, alongside undescended testis, severe congenital ptosis, and suspected congenital myasthenic syndrome. Significant cardiac and thyroid abnormalities were ruled out by a thorough preoperative evaluation, and underlying muscle dysfunction was ruled out by electromyography. The primary anaesthetic challenge involved management of a potentially difficult airway due to syndromic features and developmental status. Given the patient's age, hypotonia, and the potential of problems from traditional anaesthesia, a carefully planned anaesthetic technique was adopted utilising ProSeal Laryngeal Mask Airway (LMA) and Total Intravenous Anaesthesia (TIVA) with propofol infusion was titrated to maintain spontaneous ventilation, accompanied by caudal block using 0.2% bupivacaine for analgesia. The 90-minute surgery was uneventful with steady haemodynamics and complete recovery of patient's protective airway reflexes. This case highlights the necessity of rigorous preoperative screening, anticipation of many anaesthetic problems, and tailored perioperative management techniques in attaining optimal outcomes in children with uncommon genetic diseases.

Keywords: Chromosome deletion, Developmental disabilities, General, Orchiopexy

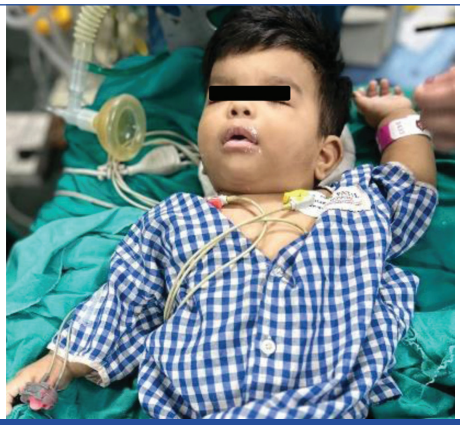
CASE REPORT

A 12-month-old male child, weighing 8.3 kg, conceived through in-vitro fertilisation and born to non-consanguineous parents via caesarean section, was admitted for orchidopexy. At nine months of age, the child had not achieved developmental milestones such as head holding, rolling over, or sitting with support. Concerned about the delayed milestones, the parents consulted a paediatrician, who diagnosed the child with intellectual and developmental disability. Subsequent genetic analysis revealed a heterozygous micro deletion in chromosome 9q13-q21.33, involving several genes associated with neurodevelopmental delay.

On examination, the child exhibited profound hypotonia and distinctive craniofacial dysmorphic features [Table/Fig-1], including microcephaly, midfacial flattening, sparse medial eyebrows, hypertelorism, and blepharophimosis. He was diagnosed with 9q13-q21.33 microdeletion syndrome, associated with undescended testis, severe congenital ptosis, and a suspected congenital myasthenic syndrome. Functional thyroid abnormalities were ruled out, and electromyography was performed to exclude underlying muscle dysfunction. Preoperative investigations, including a detailed cardiac evaluation, revealed no significant abnormalities. Due to age, lack of cooperation, and developmental delay, assessment of Mallampati classification is not possible; the airway was considered potentially difficult based on syndromic/developmental status alongside other clinical features.

Following is the genetic study report of the child showing deleted genes which are clinically significant and non-significant [Table/Fig-2,3].

The child was electively accepted for surgery under the American Society of Anaesthesiologists (ASA) Physical Status Classification III, and high-risk informed consent was obtained from the parents in view of the underlying congenital condition. Standard Nil Per Oral (NPO) guidelines were followed. On the day of surgery, the patient was taken to the operating room with a 22-gauge intravenous cannula already secured in the ward. Intravenous fluids (Dextrose 1% in Ringer's Lactate) were administered at maintenance rates.



[Table/Fig-1]: a) Patient with the craniofacial dysmorphic feature.

Gene	Phenotype	Inheritance
TMC1	Deafness, autosomal dominant 36 (OMIM#606705)	Autosomal dominant
TRPM6	Hypomagnesaemia 1, intestinal (OMIM#602014)	Autosomal recessive
VPS13A	Choreoacanthocytosis (OMIM#200150)	Autosomal recessive
CEP78	Cone-rod dystrophy and hearing loss (OMIM#617236)	Autosomal recessive
GNAQ	Capillary malformations, congenital, 1, somatic, mosaic (OMIM#163000)	Autosomal dominant
PSAT1	Phosphoserine aminotransferase deficiency (OMIM#610992)	Autosomal recessive
RORB	Epilepsy, idiopathic generalised, susceptibility to, 15 (OMIM#618357)	Autosomal dominant

[Table/Fig-2]: Deleted genes which are clinically significant.

TLE1	FRMD3	AL137018.1	FOXB2
TLE4	RASEF	AL583829.1	GCNT1
VPS13A-AS1	SPATA31D1	ALDH1A1	GDA
ZFAND5	SPATA31D3	ANXA1	GNA14
AL158154.3	ABHD17B	C9ORF40	LINC01504
AL162726.3	CARNMT1	C9ORF57	NMRK1
PRUNE2	CARNMT1-AS1	C9ORF85	OSTF1
CEMIP2	PCA3	PCSK5	RFK
	RORB-AS1		

[Table/Fig-3]: Genes which are deleted but not clinically significant.

Standard monitoring, including electrocardiography, pulse oximetry, non-invasive blood pressure, and axillary dermal temperature, was established. The patient's baseline vital signs were stable, with an SpO_2 of 100%, heart rate of 94 beats per minute, non-invasive blood pressure of 96/60 mmHg, and a temperature of 36.4°C.

Premedication included intravenous glycopyrrolate 4 µg/kg, fentanyl 1 µg/kg, and ketamine 1 mg/kg. Anaesthesia was induced with intravenous propofol 1 mg/kg, administered slowly in titrated doses to maintain spontaneous ventilation. A ProSeal LMA size 1.5 was inserted carefully with the head maintained in a neutral position to avoid injury to the jaw and teeth [Table/Fig-4]. Gastric aspiration was performed through the gastric port using a size 8 Ryle's tube. Anaesthesia was maintained with a continuous propofol infusion (2.5 mg/mL at 3 mL/h) along with a 50:50 oxygen and air mixture, allowing for spontaneous and occasional assisted ventilation to maintain an End-tidal CO_2 ($EtCO_2$) of 35-38 mmHg. A caudal block was administered using 8 mL of 0.2% bupivacaine for intraoperative analgesia.

[Table/Fig-4]: Patient being ventilated with proSeal LMA.

The procedure lasted approximately 90 minutes. Intravenous paracetamol 120 mg and dexamethasone 0.1 mg/kg administered Intraoperatively. Vitals remained stable and within preoperative limits throughout the surgery. A total of 70 mL of DRL was infused intraoperatively, and the estimated blood loss was approximately 30 mL. At the end of the procedure, the propofol infusion was discontinued (total volume infused: 4.3 mL, equivalent to 10.75 mg), and the LMA was removed after confirming adequate spontaneous ventilation and the return of protective airway reflexes.

DISCUSSION

Cytogenetically detectable interstitial deletions of 9q have been rarely reported in literature, hence do not define distinctive phenotype [1]. Boudry-Labis E et al., have recently reported nine individuals with a new microdeletion at 9q21.13 presenting with speech delay, mental impairment, epilepsy, and distinctive facial traits [2]. Moreover, Bartnik M et al., have documented a single case with epilepsy, eyelid myoclonus, generalised tonic-clonic seizures, and autism [3]. Baglietto MG et al., have also documented a case of idiopathic partial epilepsy with modest intellectual disability [4].

Among the 40 genes and transcripts specifically deleted in our patient, three previously reported Mendelian disease genes (NTRK2, SECISBP2, and AUH) are included. An illness combining mood disorders, autism, Alzheimer's disease, and developmental delays is linked to mutations in NTRK2. Type 1 methylglutaconic aciduria and an aberrant thyroid metabolism disorder are autosomal recessive disorders linked to mutations in the AUH and SECIBP2 genes, respectively [4]. The gene known as SBP2, located on chromosome 9q22.2, majority of the 25 known human selenoproteins are synthesised at a lower rate when this gene is mutated because it causes abnormalities in the incorporation of selenocysteine [5,6]. Axial muscular dystrophy, photosensitivity, immunodeficiencies, azoospermia, and aberrant thyroid function have all been correlated to deficits in selenoproteins [7]. As a result, people who have 9q deletions exhibit a variety of clinical traits with few similarities.

Therefore, important findings from this deletion include global developmental delay, aberrant thyroid function, epilepsy autistic behaviour/behavioural disorders, and dysmorphic features, including hypertelorism, long and smooth philtrum, open and huge mouth, and thin upper lips.

Chromosome 9p21 deletion presents with multiple anaesthetic challenges including: 1) difficult airway; 2) congenital hypothyroidism; 3) hypotonia; and 4) anaesthetic implications unique to paediatric age group. Facial dysmorphia creates major difficulty for airway management in children. Functional thyroid abnormalities, predominantly hypothyroidism causing macroglossia, contribute to difficult airway.

Since literature regarding anaesthetic management in this syndromic involvement is scarce, a carefully planned anaesthetic approach was adopted to avoid anticipated complications [8]. Compared to endotracheal intubation, ProSeal LMA is less invasive, can be inserted more quickly and easily, avoiding the need for muscle relaxants and inhalational agent, for deep anaesthesia. For short-duration surgical procedures in a one-year-old, this reduces patient stress alongside faster recovery time. Thus, in this case, airway was successfully secured with proseal LMA [9]. Chromosome 9p22 deletion syndrome has been associated with hypotonia [10], however no proven evidence of association with the gene 9q deletion has been reported. This poses unique challenges in anaesthetic management, making TIVA techniques more preferable in this patient with propensity to cause hyperkalaemia and malignant hyperthermia from use of inhaled anaesthetic agents. Considering anaesthetic action potentially causing respiratory depression, propofol infusion was titrated to keep spontaneous respiration intact. In view of reducing anaesthesia requirements, caudal anaesthesia was given.

CONCLUSION(S)

This case highlights the perioperative management of a child with the rare 9q21.13 deletion syndrome. Anticipated challenges included syndromic difficult airway, profound hypotonia with suspected congenital myasthenic syndrome and developmental delay. A ProSeal LMA with spontaneous ventilation, propofol-based TIVA, and caudal epidural analgesia avoided muscle relaxants and inhalational agents, limiting respiratory and neuromuscular risks. Haemodynamic stability, preserved airway reflexes, and smooth recovery highlight how tailored, minimally invasive techniques can optimise safety in such high-risk children. The successful anaesthetic course highlights the efficacy of ProSeal LMA and TIA in this high-risk paediatric patient.

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