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Anaesthetic Management of Nablus Mask Like Facial Syndrome: A Difficult Airway Case

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Abstract: ***Introduction: Nablus mask-like facial syndrome (NMLFS) is an extremely rare genetic disorder characterized by distinctive craniofacial dysmorphism and multiple congenital anomalies. From an anaesthetic perspective, the presence of micro-retrognathia, restricted mouth opening, macroglossia, restricted temporomandibular and cervical mobility, and altered upper airway anatomy places affected children at high risk for difficult mask ventilation and tracheal intubation. Published literature describing perioperative airway management in NMLFS is limited, making individual case reports valuable for guiding clinical practice. Case Description: We report the case of a two-year-old male child with genetically confirmed NMLFS who presented for elective left blepharoptosis correction. Preoperative airway evaluation revealed multiple predictors of difficult airway, including Glottic visualisation and restricted mouth opening, macroglossia, high-arched palate, short neck, and restricted cervical mobility. Anticipating airway difficulty, a comprehensive, stepwise airway management plan was implemented. Following inhalational induction with maintenance of spontaneous ventilation, both direct and video-assisted laryngoscopy failed due to poor glottic visualization. Subsequent nasal fibreoptic intubation attempts were unsuccessful. The airway was ultimately secured using a supraglottic airway–guided fibreoptic intubation technique, which allowed effective ventilation and atraumatic tracheal intubation without hypoxic compromise. Conclusion: This case highlights the significant anaesthetic challenges associated with NMLFS and underscores the importance of meticulous preoperative assessment, anticipation of difficulty, and early escalation to rescue airway techniques. Supraglottic airway–assisted fibreoptic intubation proved to be a safe and effective strategy following failed conventional approaches. Reporting such cases adds to the limited evidence base and reinforces key principles of paediatric difficult airway management aimed at optimizing perioperative safety in rare craniofacial syndromes.***

Keywords: Nablus mask-like facial syndrome, difficult airway, fibreoptic intubation, supraglottic airway, paediatric anaesthesia

1. Introduction

Difficult airway management remains one of the most critical responsibilities of the anaesthesiologist, particularly in paediatric patients with congenital syndromes. A difficult airway is defined as a clinical situation in which a trained anaesthetist experiences difficulty with face mask ventilation, tracheal intubation, or both. Syndromic children pose a proportionately higher risk due to the coexistence of craniofacial dysmorphism, restricted cervical mobility, altered airway anatomy, and limited physiological reserve, making airway control unpredictable and potentially life-threatening.[1,2]

Nablus mask-like facial syndrome (NMLFS) is an extremely rare genetic disorder characterized by a distinctive "mask-like" facial appearance and multiple congenital anomalies. First described in association with a microdeletion involving chromosome 8q22.1, the syndrome is marked by tight, glistening facial skin, blepharophimosis, sparse eyebrows, flat and broad nasal bridge, long philtrum, abnormal ear morphology, maxillary hypoplasia, and retrognathia.[3,4] Additional systemic manifestations include camptodactyly, joint contractures, cryptorchidism, developmental delay and varying degrees of neurodevelopmental impairment.[3]

From an anaesthetic perspective, the constellation of craniofacial abnormalities—particularly micrognathia, restricted temporomandibular joint mobility, limited neck extension, macroglossia, and high-arched palate—places patients with NMLFS at exceptionally high risk for difficult mask ventilation and failed direct laryngoscopy. Furthermore, the rarity of the syndrome means that most anaesthesiologists have little or no prior experience managing such airways, increasing the importance of meticulous planning, availability of advanced airway devices, and the presence of senior expertise.[5]

Genetically, NMLFS is associated with variable-sized deletions in the 8q22.1 region. Jamuar et al. reported that partial or complete deletion of the long non-coding RNA gene LINC00535 may represent a critical region; however, phenotypic variability suggests that this deletion may be necessary but not sufficient to produce the full clinical syndrome.[6] Jain et al. further demonstrated that deletion of this region without the characteristic facial phenotype is possible, underscoring the genetic heterogeneity of the disorder.[7]

Given the paucity of literature describing anaesthetic management in NMLFS, each case contributes valuable insights into airway strategies and perioperative safety. We report a case of a two-year-old child with confirmed NMLFS undergoing corrective surgery for blepharoptosis, highlighting the anticipated difficult airway and the successful use of a stepwise, rescue-oriented airway management approach.

2. Case Description

A two-year-old male child, born to non-consanguineous parents and conceived via intrauterine insemination using

self-gametes, was admitted for elective left blepharoptosis correction. Antenatal ultrasonography had revealed intrauterine growth restriction and mild micro-retrognathia. The child was delivered at term by lower-segment caesarean section for failed progression of labour, with a birth weight of 2.25 kg and immediate cry at birth. He required a one-day neonatal intensive care unit stay for evaluation of congenital anomalies.

Postnatally, the child exhibited feeding difficulties, short stature, joint contractures involving the knees, elbows, and fingers, and global developmental delay. Facial dysmorphism was striking, with a contracted puckered facial appearance, pinched mouth, restricted mouth opening, micro-retrognathia, poor scalp hair, absent eyebrows and eyelashes, blepharophimosis, flat broad nasal bridge, short columella, long philtrum, high-arched palate, and low-set dysplastic ears with absent external auditory meatus. Additional anomalies included hypospadias, bilateral undescended testes, inguinoscrotal hernia, umbilical hernia, scissoring of the lower limbs, and episodic bluish discoloration of the lower face during crying. Severe-to-profound hearing loss was documented..

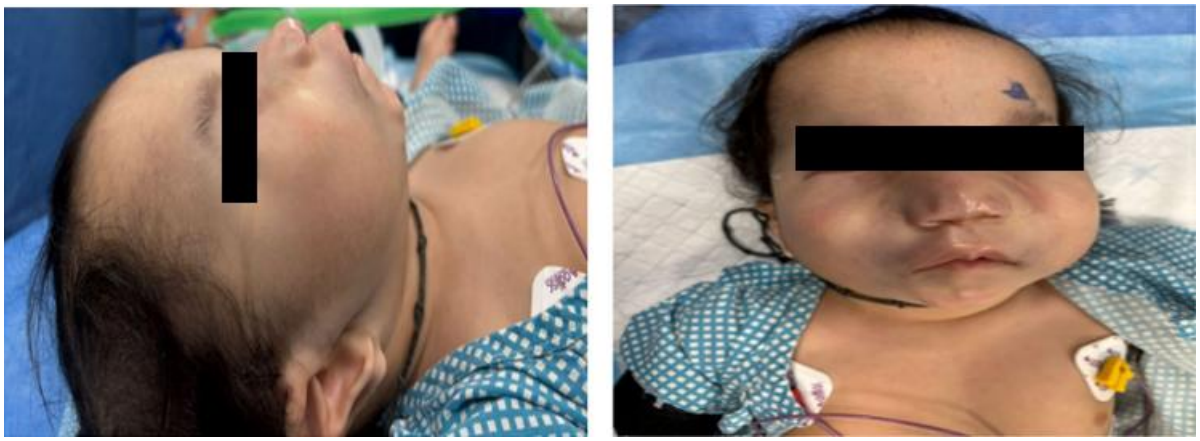

Figure 1: Clinical photographs of the child with Nablus mask-like facial syndrome demonstrating characteristic craniofacial dysmorphism. The lateral view (left) shows micro-retrognathia, short neck, and tight, smooth facial skin, while the frontal view (right) highlights a mask-like facial appearance with blepharophimosis, flat and broad nasal bridge, long philtrum, small pinched mouth, and reduced facial expression.

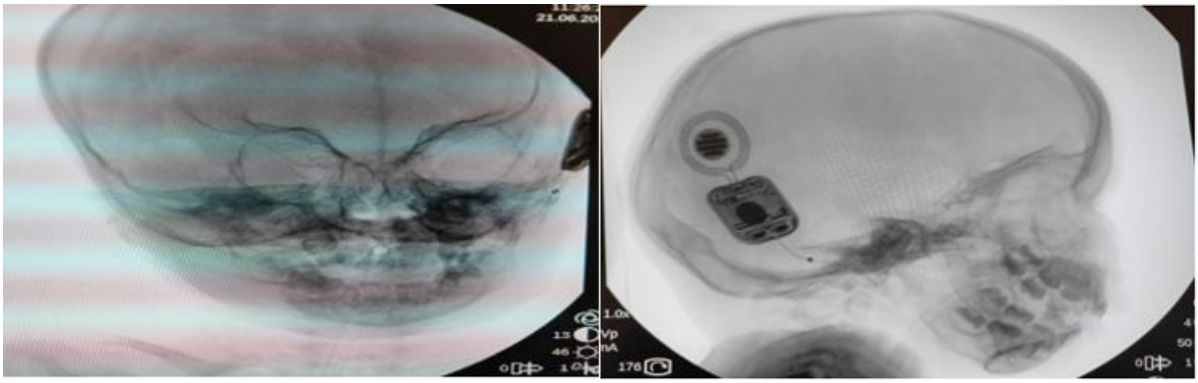

Figure 2: Lateral skull radiographs demonstrating craniofacial abnormalities in the child with Nablus mask-like facial syndrome. The images show microcephaly with altered cranial contour, mandibular hypoplasia with retrognathia, and reduced lower facial projection, consistent with the characteristic craniofacial dysmorphism associated with this syndrome

Developmental milestones were delayed, with neck control achieved at four months and brief sitting with support by eleven months. On admission, the child weighed 8.8 kg and measured 78 cm in height. Neurological examination revealed increased tone in all four limbs with preserved muscle strength. Cardiopulmonary examination was unremarkable.

Airway assessment suggested a potentially difficult airway, with macroglossia, restricted mouth opening, high-arched palate, restricted temporomandibular joint movement, short neck, and reduced cervical mobility. Based on clinical features and prior genetic evaluation, a diagnosis of Nablus mask-like facial syndrome was confirmed.

3. Investigations

Baseline laboratory investigations showed haemoglobin of 12 g/dL, total leukocyte count of 10,300/mm³, platelet count of 538,000/mm³, normal coagulation profile (PT/INR 13.6/0.94), and serum creatinine of 0.39 mg/dL. Two-dimensional echocardiography demonstrated situs solitus with normal cardiac anatomy, intact interventricular septum, patent foramen ovale with left-to-right shunt, normal biventricular function, and no evidence of ductus arteriosus or aortic coarctation. Chest radiograph was unremarkable.

4. Management

In view of the anticipated difficult airway, a structured airway management plan was formulated preoperatively, with emphasis on maintaining oxygenation and minimizing airway trauma. A complete difficult airway setup was prepared in advance, including age-appropriate face masks, oral and nasal airways, laryngeal mask airways, cuffed and uncuffed endotracheal tubes, stylet, paediatric bougie, Miller and Macintosh blades, video laryngoscope with Miller blade, paediatric fibreoptic bronchoscope (2.8 mm), and suction apparatus. A senior paediatric anaesthesiologist was present throughout induction and airway manipulation.

To reduce preoperative anxiety while preserving airway reflexes, the child was premedicated in the preoperative holding area with 5% midazolam nasal spray administered in the presence of the mother. The operating room ambient temperature was maintained at approximately 24 °C to reduce the risk of perioperative hypothermia, Induction was initiated using an inhalational technique with sevoflurane, oxygen and air mixture via a Jackson–Rees circuit, allowing maintenance of spontaneous respiration with saturation probe connected to patient. After patient achieved adequate depth of anaesthesia and calm, rest of all standard ASA monitors were attached. Mask ventilation was confirmed under inhalational anaesthesia. Intravenous access was secured with a 24G cannula after adequate depth of anaesthesia was achieved.

Following induction, intravenous midazolam 0.4 mg, glycopyrrolate 80 µg, and fentanyl 10 µg were administered, followed by propofol 20 mg. After confirming effective mask ventilation, atracurium 5 mg was given to facilitate airway instrumentation. The child was ventilated with a face mask for three minutes prior to laryngoscopy.

Video laryngoscopy using a Miller blade revealed a large tongue, restricted mouth opening, limited mandibular elevation, and restricted cervical spine mobility, permitting visualization of only the epiglottic tip despite optimal external laryngeal manipulation.

An initial attempt at tracheal intubation was made using a 3.5mm cuffed endotracheal tube by blind advancement beneath the epiglottis; however, the attempt was unsuccessful. Ventilation was maintained with bag and mask between attempts with oxygen saturation remaining acceptable. A further attempt at tracheal intubation using a 3.5 mm cuffed endotracheal tube mounted on a stylet, which was unsuccessful. Adequate mask ventilation was re-established. Thereafter, nasal fibreoptic-guided intubation was attempted using a 2.8 mm fibreoptic bronchoscope without a suction port, with a 3.5 mm cuffed endotracheal tube preloaded over the scope, but the endotracheal tube could not be advanced beyond the glottis. Oxygenation was maintained throughout the procedure.

Recognizing the limitations of further laryngoscopy attempts, the airway strategy was escalated to a rescue technique. A 3.5-mm cuffed endotracheal tube was first tested ex vivo through the i-gel to ensure that the supraglottic airway could be removed easily after successful intubation. A size 1.5 I-gel supraglottic airway device was inserted, achieving effective ventilation. Fibreoptic bronchoscopy was then performed through the I-gel, and a 3.5 mm uncuffed endotracheal tube was successfully advanced into the trachea under direct visualization. The supraglottic airway was removed by railroading over a 3 mm cuffed endotracheal tube.

After successful ventilation with I-gel, the airway strategy was escalated to secure definitive airway through supraglottic device as child had markedly limited mouth opening and distorted intraoral anatomy, resulting in a restricted intraoral space. Definitive tracheal intubation through I-gel was planned due to potential displacement resulting in loss of airway.

Following successful placement of the endotracheal tube through the supraglottic airway device, correct tracheal placement was confirmed under direct visualization of the carina with optimal positioning of the tip of the endotracheal tube. Tube position was further confirmed by bilateral chest auscultation and continuous capnography. The supraglottic airway device was then carefully removed while maintaining stabilization of the tracheal tube using a size 3 endotracheal tube as an obturator, thereby preventing tube displacement. After removal of the supraglottic airway device, correct endotracheal tube position was reconfirmed and the tube was securely fixed.

Anaesthesia was maintained uneventfully, and surgery proceeded after ensuring stable ventilation and hemodynamic. This stepwise, escalation-based approach allowed safe airway control while avoiding repeated traumatic attempts in a child with a high-risk syndromic airway.

5. Extubation

Extubation was planned as a high-risk extubation due to anticipated difficulty in re-establishing the airway. The child was allowed to regain full consciousness with adequate spontaneous ventilation, adequate tidal volume, respiratory efforts and stable hemodynamic parameters. Complete neuromuscular blockade was confirmed prior to extubation. Adequate oxygenation and airway patency were ensured, and the patient was thoroughly suctioned. Airway oedema was ruled out clinically.

A difficult airway cart including appropriately sized endotracheal tubes, supraglottic airway devices, video laryngoscope, and fibreoptic bronchoscope was kept readily available and Senior paediatric anaesthesiologists were present during extubation.

Following extubation nursing in recovery position, supplemental oxygen was administered via face mask, and the child was closely monitored for any signs of airway obstruction, stridor, desaturation, or respiratory distress. The patient maintained adequate spontaneous ventilation with stable oxygen saturation and no immediate post-extubation airway complications were observed.

A few months after the initial procedure, the child was readmitted for a second surgical intervention. In view of the persistent craniofacial anatomical variation and the

anticipated difficult airway, a similar airway management strategy was planned as used during the first surgery. The previous anaesthetic record was reviewed, and all necessary difficult airway equipment was kept readily available. Following adequate preoxygenation, airway management was performed using the previously successful technique and rest of anaesthetic course was uneventful.

6. Discussion

This case underscores the profound anaesthetic challenges posed by rare craniofacial syndromes such as NMLFS. The defining facial characteristics—mask-like appearance, micro-retrognathia, restricted jaw mobility, limited mouth opening and cervical stiffness—create a composite airway that is both anatomically distorted and functionally limited. Such features are well-recognized predictors of difficult laryngoscopy and intubation in paediatric practice.[8,9]

In the present case, failure of both direct and video-assisted laryngoscopy suboptimal positioning due to limited cervical mobility and external laryngeal manipulation highlights the limitations of conventional laryngoscopy techniques in syndromic airways. Visualization of only the epiglottic tip is a well-recognized finding in children with mandibular hypoplasia and midface retrusion, as reported in previous paediatric airway studies.[9] These anatomical constraints emphasize that video laryngoscopy, although valuable, does not universally guarantee improved glottic exposure in all difficult airways.

A key strength of airway management in this case was the early recognition of laryngoscopy failure followed by a strategic decision to limit further intubation attempts and proceed with an alternative airway approach. Multiple laryngoscopy attempts are known to increase airway trauma, oedema, bleeding, and the risk of complete airway obstruction, particularly in children.[10] The decision to promptly escalate to a supraglottic airway–based rescue strategy aligns with contemporary paediatric difficult airway algorithms, which emphasize oxygenation over intubation and advocate early use of alternative techniques when initial attempts fail.[10, 11]

The successful use of a supraglottic airway as a conduit for fibreoptic-guided tracheal intubation further reinforces the utility of this technique in anticipated difficult paediatric airways. Supraglottic devices provide a stable, ventilated airway while allowing controlled fibreoptic visualization and atraumatic tube placement. Clinical studies and expert consensus support this approach as both effective and safe, particularly when anatomical distortion precludes direct passage of an endotracheal tube.[12] In our patient, the i-gel facilitated uninterrupted ventilation and served as a reliable conduit for fibreoptic intubation, ultimately enabling secure airway control without hypoxic compromise.

Maintaining spontaneous ventilation during induction, administering neuromuscular blockade only after confirming effective mask ventilation, and ensuring the presence of senior expertise were additional factors that contributed to the favourable outcome. These measures are strongly recommended in syndromic paediatric airways, where loss of airway control can be catastrophic.[5]

The rarity of NMLFS means that perioperative experience is limited to isolated case reports, making standardized airway algorithms difficult to establish. However, this case reinforces several key principles: anticipation of difficulty, preparation of a comprehensive difficult airway cart, preservation of spontaneous ventilation until airway security is assured, and early transition to alternative airway techniques rather than repeated traumatic laryngoscopy attempts.[13]

Given the associated developmental delay and potential multisystem involvement in NMLFS, a multidisciplinary perioperative approach involving anaesthesiologists, geneticists, surgeons, and paediatricians is essential. As more cases are reported, collective experience will help refine airway management strategies and improve perioperative outcomes in this rare population.

7. Conclusion

Nablus mask-like facial syndrome is an exceptionally rare genetic condition with distinctive craniofacial features that pose significant anaesthetic challenges, particularly in airway management. This case illustrates the complexity of securing the airway in a syndromic paediatric patient with micro-retrognathia, limited mouth opening, restricted temporomandibular and cervical mobility, and altered upper airway anatomy. Despite the failure of conventional and video-assisted laryngoscopy, a structured, stepwise approach incorporating supraglottic airway-assisted fibreoptic intubation enabled safe airway control without hypoxic compromise.

Anticipation of a difficult airway, meticulous preoperative planning, maintenance of spontaneous ventilation during induction, and immediate availability of advanced airway devices are critical to successful outcomes in such cases. Equally important is the presence of experienced personnel capable of rapidly transitioning to alternative airway techniques when initial attempts fail.

As literature on anaesthetic management in NMLFS remains sparse, reporting such cases is vital to expanding collective knowledge and guiding future practice. Awareness of this syndrome and its airway implications can help anaesthesiologists anticipate challenges, avoid repeated traumatic attempts, and adopt safer airway strategies. Ultimately, individualized planning and adherence to difficult airway principles remain the cornerstone of anaesthetic care in rare craniofacial syndromes.

Conflict of Interest
Nil declared by the authors

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