DOI: 1021088/ijpen.2454.9126.12226.4

International Journal of Pediatric Nursing
Volume 12 Number 2, May – August 2026

CASE REPORT

Craniosynostosis with Midfacial Hypoplasia and Bilateral Choanal Atresia Presenting with Respiratory Distress and Acute Kidney Injury in an Infant: A Case Report

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HOW TO CITE THIS ARTICLE:

Jayavel M, Rashmi Singh, Joyce Joseph et. al, Craniosynostosis with Midfacial Hypoplasia and Bilateral Choanal Atresia Presenting with Respiratory Distress and Acute Kidney Injury in an Infant: A Case Report. Int J Pediatr Nurs. 2026; 12(2): 99–103.

ABSTRACT

Background: Craniosynostosis is a congenital condition characterized by premature fusion of cranial sutures, leading to abnormal skull growth and potential neurological and respiratory complications. It may be associated with syndromic conditions such as Crouzon syndrome.

Case Presentation: A 6-month-old female infant presented with cough, cold, fever, and lethargy. She had a history of prolonged neonatal intensive care unit (NICU) stay and delayed cry at birth. On examination, the child was undernourished, lethargic, and had abnormal craniofacial features, including frontal bossing, midfacial hypoplasia, depressed nasal bridge, short neck, and limb deformities (syndactyly and oligodactyly). Respiratory distress with stridor was noted. The patient was diagnosed with craniosynostosis syndrome with midfacial hypoplasia, bilateral choanal atresia, and stage 3 acute kidney injury.

Management and Outcome: The child received supportive care including oxygen therapy, fluid management, and monitoring in a pediatric intensive care unit (PICU). Nursing care focused on airway management, monitoring vital parameters, and maintaining hygiene and nutrition.

Conclusion: Early recognition of craniosynostosis and associated syndromic features is essential for timely intervention. Multidisciplinary management plays a key role in improving outcomes.

KEYWORDS

• Craniosynostosis • Choanal Atresia • Midfacial Hypoplasia • Infant • Respiratory Distress • Picu Care

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➢ **Received:** 15-04-2026 ➢ **Accepted:** 20-05-2026

INTRODUCTION

Craniosynostosis is a congenital craniofacial disorder characterized by the premature fusion of one or more cranial sutures, resulting in restricted skull growth perpendicular to the affected suture and compensatory overgrowth in other areas. This leads to abnormal head shape and, in some cases, increased intracranial pressure, which may affect brain development if left untreated. The condition may occur as an isolated anomaly or as part of syndromic associations such as Crouzon syndrome, Apert syndrome, or Carpenter syndrome.

Associated craniofacial abnormalities, including midfacial hypoplasia and choanal atresia, can contribute to upper airway obstruction, feeding difficulties, and recurrent respiratory distress in infants. These anomalies may further compromise oxygenation and overall growth. Early recognition through clinical examination and imaging, followed by timely surgical intervention and multidisciplinary care involving pediatricians, neurosurgeons, and otolaryngologists, is crucial to prevent complications such as neurodevelopmental delay, visual impairment, and respiratory failure.

CASE PRESENTATION

A 6-month-old female infant was admitted to the Pediatric Intensive Care Unit with complaints of cough, cold, and fever for 7 days, associated with lethargy. The cough was non-productive and accompanied by post-tussive vomiting. The fever was low grade, with a maximum recorded temperature of approximately 100°F, and was temporarily relieved with medications. The child was also noted to be less active than usual during this period.

The infant was apparently well 7 days prior to admission, after which she developed the above symptoms. Due to worsening lethargy and persistent respiratory symptoms, she was initially managed at a local hospital where she received oxygen therapy via nasal prongs and intravenous fluids before being referred for further management.

The child had a significant neonatal history, including delayed cry at birth and admission to the neonatal intensive care unit (NICU) for 25 days. During that period, she required mechanical ventilation for 3 days and oxygen support for 17 days, indicating early respiratory compromise.

Antenatal history was uneventful, with no reported maternal illness, drug exposure, or complications during pregnancy. The child was born at term via normal vaginal delivery with a birth weight of 3.2 kg.

Immunization status was incomplete for age. Developmental milestones could not be adequately assessed due to the child's current illness and underlying condition.

The child belonged to a low socioeconomic background. There was no significant family history of congenital anomalies, genetic disorders, or chronic illnesses.

Clinical Examination

On examination, the infant was lethargic, irritable, and undernourished, with features suggestive of poor general condition. Vital signs revealed tachycardia (pulse: 140 beats/min), respiratory rate of 30 breaths/min, and hypotension (blood pressure: 71/40 mmHg), indicating hemodynamic instability.

Anthropometric assessment showed weight significantly below expected for age, suggestive of undernutrition.

Detailed physical examination revealed multiple craniofacial abnormalities, including frontal bossing, asymmetrical head shape, and premature closure of cranial sutures. A depressed nasal bridge, midfacial hypoplasia, and short neck were also noted, consistent with syndromic craniosynostosis.

Respiratory examination revealed stridor and reduced air entry bilaterally, suggestive of upper airway obstruction, likely secondary to midfacial hypoplasia and possible choanal atresia.

Musculoskeletal examination demonstrated multiple limb anomalies, including syndactyly, oligodactyly, and rocker-bottom feet, indicating an associated congenital syndrome.

DIAGNOSIS

The patient was diagnosed with:

- Craniosynostosis syndrome
- Midfacial hypoplasia
- Bilateral choanal atresia
- Stage 3 acute kidney injury
- Respiratory distress

Pathophysiology

Craniosynostosis results from premature fusion of cranial sutures, leading to restricted skull growth perpendicular to the affected suture and compensatory expansion in other areas, resulting in abnormal head shape. This may increase intracranial pressure, leading to risk of brain compression and developmental delay. Associated midfacial hypoplasia causes narrowing of the nasopharyngeal airway, while bilateral choanal atresia leads to obstruction of nasal airflow. Together, these abnormalities result in respiratory distress and hypoxia.

MANAGEMENT

Supportive and Medical Management

1. Oxygen Therapy

- Administer humidified oxygen via nasal prongs, mask, or CPAP depending on severity
- Maintains adequate oxygen saturation and prevents hypoxia
- Continuous monitoring using pulse oximetry is essential

2. Intravenous (IV) Fluids

- Given to maintain hydration and electrolyte balance, especially if feeding is difficult
- Prevents dehydration due to respiratory distress
- Careful fluid regulation is required to avoid fluid overload, particularly in cases with raised intracranial pressure

3. Supportive Management in PICU (Pediatric Intensive Care Unit)

- Continuous monitoring of vital signs (HR, RR, SpO_2, BP)
- Airway management: positioning (head elevation), suctioning, and readiness for intubation if needed
- Mechanical ventilation in severe respiratory distress or airway obstruction
- Monitoring for signs of increased intracranial pressure (bulging fontanelle, vomiting, irritability)
- Nutritional support (NG tube feeding if oral feeding is compromised)
- Infection prevention and management (aseptic care, antibiotics if indicated)

Nursing Management (Care Plan)

1. Ineffective airway clearance related to upper airway obstruction secondary to Midfacial hypoplasia and Choanal atresia as evidenced by respiratory distress, nasal obstruction, use of accessory muscles

Goals:

- Child will maintain a patent airway
- SpO_2 > 94%

Interventions:

- Position in semi-Fowler's position
- Administer oxygen therapy as prescribed
- Gentle suctioning of secretions
- Monitor respiratory rate, effort, and SpO_2
- Keep emergency airway equipment ready

Evaluation:

- Airway remains clear, no distress, SpO_2 maintained

2. Impaired gas exchange related to airway obstruction and hypoxia as evidenced by cyanosis, low oxygen saturation, irritability

Goals:

- Maintain adequate oxygenation
- ABG values within normal range

Interventions:

- Continuous pulse oximetry monitoring
- Administer humidified oxygen
- Assist with ventilation if required (CPAP/ventilator)
- Monitor for signs of hypoxia (restlessness, cyanosis)

Evaluation:

Improved oxygen saturation and clinical condition

3. Imbalanced nutrition: less than body requirements related to difficulty in feeding due to respiratory distress as evidenced by poor feeding, weight loss/inadequate weight gain.

Goals:

- Child will achieve adequate nutritional intake
- Maintain appropriate weight for age

Interventions:

- Provide small, frequent feeds
- Use NG tube feeding if oral feeding is difficult
- Monitor daily weight
- Collaborate with dietician

Evaluation:

- Improved feeding and weight gain

4. Acute pain related to surgical intervention or increased intracranial pressure in Craniosynostosis as evidenced by crying, irritability, facial grimacing

Goals:

- Pain will be reduced or relieved

Interventions:

- Assess pain using appropriate scale
- Administer analgesics as prescribed
- Provide comfort measures (positioning, swaddling)
- Reduce environmental stimuli

Evaluation:

- Child appears comfortable, reduced pain signs

5. Risk for Infection related to invasive procedures and compromised airway

Goals:

- Prevent/control infection
- Interventions:
- Maintain strict aseptic technique
- Monitor temperature and lab values
- Administer antibiotics as prescribed
- Ensure proper hygiene and handwashing
- Evaluation:
- No signs of infection/infection controlled

DISCUSSION

Craniosynostosis is a complex congenital craniofacial disorder that often presents with varying degrees of multisystem involvement, particularly in syndromic cases. The premature fusion of cranial sutures not only affects skull shape but may also lead to increased intracranial pressure and impaired neurodevelopment if not managed early.

Similar findings have been reported in previous studies highlighting the association between craniosynostosis and airway compromise in syndromic cases.

In the present case, the coexistence of Midfacial hypoplasia and bilateral Choanal atresia played a significant role in causing upper airway obstruction and subsequent respiratory distress. Midfacial hypoplasia results in reduced nasopharyngeal space, while choanal atresia further compromises nasal airflow, making the infant highly dependent on oral breathing. This combination substantially increases the risk of hypoxia and respiratory failure, especially during feeding or sleep.

The presence of associated limb anomalies in this case raises suspicion of a syndromic form of craniosynostosis, possibly overlapping with conditions such as Crouzon syndrome or Carpenter syndrome. Syndromic craniosynostosis is often linked with genetic mutations (commonly involving FGFR genes) and may present with additional systemic manifestations, necessitating comprehensive genetic evaluation and counseling.

Respiratory compromise remains one of the most critical and life-threatening concerns in such patients due to structural airway abnormalities. The requirement for Pediatric Intensive Care Unit (PICU) support in this case reflects the severity of airway obstruction and the need for continuous monitoring, oxygen supplementation, and possible ventilatory assistance. Early airway stabilization is essential to prevent complications such as prolonged hypoxia and its neurological consequences.

Furthermore, the development of acute kidney injury added complexity to the clinical course, likely secondary to hypoxia, poor perfusion, or systemic stress. This highlights the importance of vigilant monitoring of organ function and timely supportive management in critically ill infants.

Overall, this case underscores the importance of early diagnosis, prompt recognition of associated anomalies, and a multidisciplinary approach involving paediatricians, neurosurgeons, otolaryngologists, geneticists, and nursing staff. Timely surgical intervention combined with optimal supportive care can significantly improve outcomes and reduce long-term morbidity.

CONCLUSION

Craniosynostosis with associated airway anomalies represents a potentially life-threatening condition requiring early recognition and prompt intervention. This case highlights the significant impact of coexisting Midfacial hypoplasia and Choanal atresia in precipitating severe respiratory distress in infancy.

The presence of additional anomalies, including limb defects and acute kidney injury, suggests a possible syndromic association, emphasizing the need for thorough clinical evaluation and genetic assessment. Early multidisciplinary management, including intensive supportive care and timely surgical correction, plays a crucial role in improving survival and long-term neurodevelopmental outcomes.

This case reinforces the importance of a holistic approach that integrates medical, surgical, and nursing care to effectively manage complex congenital conditions.

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