# Case Report: Anaesthetic management of an infant with Nager syndrome and a complex congenital heart defect

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## Abstract

### Introduction/Study Question

The main Nager syndrome clinical features include craniofacial, limb and musculoskeletal malformations. Complex congenital cardiac defects are rare. The objective is to present the first case of anesthetic management of a patient with Nager syndrome in Colombia, highlighting the difficult airway approach with an associated congenital heart defect.

### Methods

The clinical case is about a one month old infant, diagnosed with Nager syndrome due to micrognathia, cleft palate, hipoplastic thumbs. Transthoracic echocardiography revealed patent ductus arteriosus (PDA), ostium secundum atrial septal defect, perimembranous ventricular septal defect, leading to severe pulmonary hypertension. A multidisciplinary assessment concluded that the patient was candidate to patent ductus arteriosus ligation and, according to the evolution, to undergo primary mandibular distraction osteogenesis.

On physical examination, the infant weighed 4.6 kg, with hypoplastic mandible and glossoptosis, and signs of decompensated heart failure due to increased pulmonary blood flow. The preoperative tests were acceptable, although a persistent respiratory acidosis was noted.

Before the surgical procedure, an axillary central venous catheter was inserted in the neonatal intensive care unit (NICU). The patient fasted 6 h. Inhalatory anesthetic induction with sevoflurane was performed, a fiberoptic bronchoscope (2.8 mm) was introduced orally and tracheal intubation with a 4.0 uncuffed endotracheal tube proceeded without difficulty. Balanced general anesthesia with fentanyl 5 μg/kg and sevoflurane 0.8 MAC was maintained. Dopamine infusion was required during the procedure. Patent ductus arteriosus ligation was uneventful. The infant was transferred to the NICU. The patient remained intubated and a primary mandibular distraction osteogenesis was scheduled for a week later. Hence, inhalational induction with sevoflurane was done, a 4.0 uncuffed endotracheal tube was introduced nasally including the fiberoptic bronchoscope with adequate visualization of the glottis was accomplished. The procedure lasted 100 min.

Ten days later, the patient was extubated, after reaching a mandibular distraction of 10 mm. The patient was supported with nasopharyngeal cannula, and a tracheostomy was performed after 15 days.

### Results

Acrofacial dysostosis represents a rare malformation of unknown etiology. The anesthetic management of Nager syndrome represents a challenge as a result of the predicted difficult intubation and ventilation. Administering adequate anesthesia, so that these patients will tolerate the stimulation from bronchoscopy and intubation while maintaining spontaneous ventilation is a demanding task, being more critical in the context of increased pulmonary blood flow, where hypoxia, hypercarbia, acidosis, hypothermia and pain could increase pulmonary hypertension.

### Conclusion/Discussion

Nager syndrome airway is difficult. Fiberoptic intubation is the preferred mean to secure the airway; however, tracheostomy remains the standard for long-term management of severe airway obstruction, even though mandibular distraction osteogenesis is a surgical alternative. A multidisciplinary team should be available in the care of these patients.

## Background

Nager syndrome or acrofacial dysostosis results from aberrations in development of the first and second branchial arches and limb buds. It was first described by Nager and de Reynier in 1948, who used the term acrofacial dysostosis to distinguish the condition from mandibulofacial dysostosis.

Since then, more than 80 cases have been reported in neonates and infants. The main clinical features include craniofacial, limb and musculoskeletal malformations, with mandibular hypoplasia as a key feature, representing an important predictor of difficult airway. Other anomalies have been reported. Although speech development may be delayed due to hearing impairment, Nager syndrome does not affect a child's intelligence. Most cases appear to be sporadic, however, an autosomal dominant or an autosomal recessive pattern of inheritance have been mentioned. The pattern of inheritance remains unclear.

Of immediate concern is the precarious nature of the airway, given the degree of micro/retrognathia and trismus, causing that severe airway obstruction is the major cause of morbidity and mortality, affecting also feeding. Early intervention with tracheostomy has been emphasized. Although tracheostomy is a definitive procedure for securing the airway in children with upper airway obstruction, it is associated with potentially significant morbidity and a mortality rate.

Numerous other procedures have been advocated for the management of airway obstruction in children with craniofacial abnormalities to avoid tracheostomy or achieve decannulation.

## Objective

The purpose of this report is to present the first case of anesthetic management of a patient with Nager syndrome in Colombia, highlighting the challenges of managing a patient with difficult airway and an associated complex decompensated congenital heart defect.

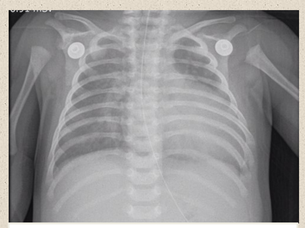
## Methods

### Clinical Case Description

The clinical case is about a one month old infant, diagnosed with Nager syndrome since the neonatal period due to these physical features: micrognathia, cleft palate, hipoplastic thumbs. The patient was referred to our institution for further evaluation because of life threatening airway obstruction. Transthoracic echocardiography revealed patent ductus arteriosus (PDA), ostium secundum atrial septal defect, perimembranous ventricular septal defect, leading to severe pulmonary hypertension. A multidisciplinary assessment including neonatology, cardiology, pediatric surgery, otorhinolaryngology, plastic surgery and anesthesiology, concluded that the patient was candidate to patent ductus arteriosus ligation and, according to the evolution, to undergo primary mandibular distraction osteogenesis, in an attempt to improve respiratory and feeding problems.

### Preoperative assessment

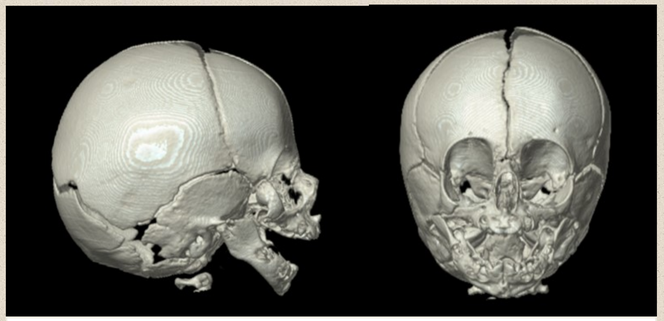
The infant weighed 4.6 kg, showed the referred typical characteristics, with remarkable difficult airway predictors: hypoplastic mandible and glossoptosis, and signs of decompensated heart failure due to increased pulmonary blood flow, despite medical treatment. The preoperative tests were acceptable, although a persistent respiratory acidosis was noted. Informed consent was obtained. Before the surgical procedure, an axillary central venous catheter was inserted in the neonatal intensive care unit (NICU).



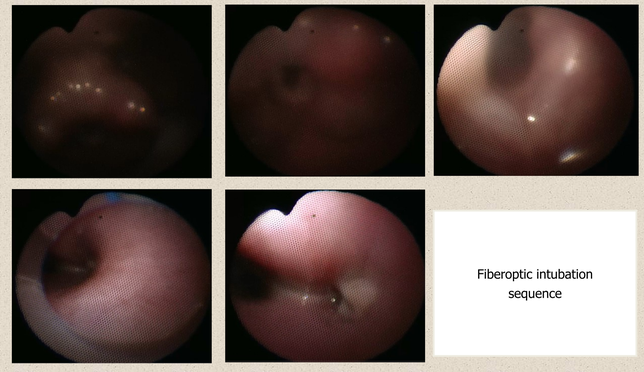
Preoperative Chest X-ray

### Intraoperative management

The difficult airway equipment was prepared. Inhalatory anesthetic induction with sevoflurane was performed, a fiberoptic bronchoscope (2.8 mm) was introduced orally and tracheal intubation with a 4.0 uncuffed endotracheal tube proceeded without difficulty. Balanced general anesthesia with fentanyl 5 μg/kg and sevoflurane 0.8 MAC was maintained. Dopamine infusion was required during the procedure. Patent ductus arteriosus ligation was uneventful and lasted 60 minutes. The infant was transferred to the NICU. The patient remained intubated, and taking into account the optimal evolution, was scheduled to undergo primary mandibular distraction osteogenesis a week later. For this procedure, inhalational induction with sevoflurane was done, a 4.0 uncuffed endotracheal tube was introduced nasally, and the fiberoptic bronchoscope was introduced through it, adequate visualization of the glottis was accomplished, and patient was intubated successfully. The procedure lasted 100 min. The patient returned intubated to the NICU.



Skull CT with 3D reconstruction

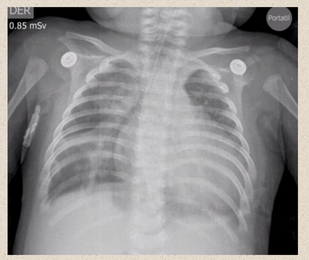


Fiberoptic intubation sequence

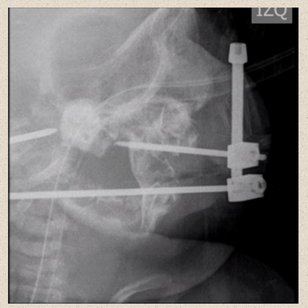
Patient images after first surgery

### Postoperative outcome

Ten days later, the patient was extubated, after reaching a mandibular distraction of 10 mm. The patient required continuous support with nasopharyngeal cannula, and a tracheostomy was performed 15 days afterwards taking into account the persistent risk of airway obstruction.



Postoperative Chest X-ray



## Results

Acrofacial dysostosis represents a rare malformation of unknown etiology, diagnosed according to clinical features and genetic testing. The anesthetic management of Nager syndrome represents a challenge as a result of the predicted difficult intubation and ventilation. Administering adequate anesthesia, so that these patients will tolerate the stimulation from bronchoscopy and intubation while maintaining spontaneous ventilation is a demanding task, being more critical in the context of increased pulmonary blood flow, where hypoxia, hypercarbia, acidosis, hypothermia and pain could increase pulmonary hypertension.

## Conclusions

Nager syndrome airway is difficult. Fiberoptic intubation is the preferred means to secure the airway; however, tracheostomy remains the standard for long-term management of severe airway obstruction, even though mandibular distraction osteogenesis is a surgical alternative. A multidisciplinary team should be available in the care of these patients.

## References

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