

# Tracheostomy Conducted in a Freeman-Sheldon Syndrome Case

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## Abstract

The Freeman-Sheldon syndrome is an autosomal dominant disease and the main features are the changes in the face with external projection of the lips (whistling position), underdeveloped nasal cartilage, narrow nostrils, microstomia and microglossia, which explains the signs of respiratory distress and swallowing problems, promoting the indication for tracheostomy. This paper reports a case of tracheostomy in a newborn baby suffering from the Freeman-Sheldon syndrome, contributing with information that can help health professionals to improve the respiratory condition in such cases and to prevent future complications with invasive mechanical ventilation in the long-term basis, justifying the indication of tracheostomy in period similar to that carried out in children without neurologic diseases.

**Keywords:** Tracheostomy; Freeman-Sheldon syndrome; Respiratory insufficiency; Child; Intensive Care Units; Neonatal; Microstomia

## Introduction

The Freeman-Sheldon syndrome was first described in 1938 as a crano-carpo-tarsal dysplasia, believed to be very rare, but seven studies approach the theme in 2013 [1-7], and, by 1990, 65 cases have been described in the literature [8, 9].

It is a syndrome of autosomal dominant inheritance; however, in some cases, it has an autosomal recessive inheritance [8]. The masculine and feminine genders are equally

affected, and, in most cases, mental retardation is not diagnosed, except when there are structural abnormalities of the nervous system [9].

The main features are bone abnormalities of the extremities, such as camptodactyly with ulnar deviation of the hands and congenital equinus and varus clubfoot. The face presents deep eyes and small palpebral fissure, inflated cheeks, with external projection of the lips (whistling position), underdeveloped nasal cartilage, narrow nostrils, microstomia and microglossia, which explains the signs of respiratory distress and swallowing problems, promoting the indication of tracheostomy (TCH) [8, 10].

Reporting the respiratory development to the scientific scope during the hospitalization of a newborn baby suffering from the Freeman-Sheldon syndrome contributes to the performance of health professionals, improving the respiratory condition in such cases and preventing future complications with invasive mechanical ventilation in the long-term basis.

The objective of this study was to describe the indication of TCH for a child suffering from the Freeman-Sheldon syndrome hospitalized in a Neonatal Intensive Care Unit.

The research designed as case report was approved by the Ethics and Research, under the protocol number 212,790; the responsible for the newborn baby signed a consent form authorizing the study. The research was conducted during the hospitalization of the volunteer in a Neonatal Intensive Care Unit at a public hospital of the state system, the city of Sao Paulo, Brazil, with the collection of data from the medical record.

## Case Report

A newborn baby (NB) of M.P.M., the child of an 18-year-old Bolivian mother who, in her first pregnancy, reported having attended ten medical visits for the prenatal care, fetal ultrasonography showed polyhydramnios, short long bones and hypoplastic left heart syndrome.

NB, masculine gender, was born at the date of 20 June, 2012, with gestational age by the Somatic Capurro method: 39 weeks 5/7, born by c-section, with the aid of lever and vertex presentation, birth weight: 2.950 kg, height at birth:

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45.5 cm, appropriate for gestational age, Apgar 5/8 in the first and fifth minutes, respectively.

The patient was born whining, and cyanotic, with heart rate: 160 beats per minute, evolving to a respiratory failure of progressive nature, during the initial moments of life, with Silverman-Anderson index of 8, having the tracheal intubation and the mechanical ventilation been indicated; the tracheal intubation was difficult due to micrognathia.

On physical examination at birth, multiple malformations were observed: low-set ears, micrognathia, tapered chest, hands and thumbs in anomalous position, bilateral simian crease and diastasis of abdominal rectus.

Four attempts of extubation were unsuccessful because the NB evolved with significant respiratory discomfort, even in noninvasive mechanical ventilation. During this period the chest X-ray has shown the formation of atelectasis, the deformity of the bone structure and the opacity of the left costophrenic sulcus. After 62 days of mechanical ventilation and the failure in attempts of extubation, the medical staff indicated the TCH.

The infant had an uneventful recovery and remained stable in oxygen therapy, with tracheal mask nebulization at 5 L/min; after 10 days of TCH, he was transferred to the pediatric intensive care unit (ICU).

## Discussion

The TCH was indicated after 2 months since the infant had remained on invasive mechanical ventilation, supporting the study conducted by Itamoto which demonstrated the intubation time of 57 days before the TCH [11].

The success of extubation in the ICUs is often associated with the clinical condition of the child, as there are few studies that demonstrate predictive indexes that allow the ventilatory weaning or the best time to conduct the TCH.

Thus, we find in the case report that narrow nostrils, microstomia and microglossia linked to the anatomical and physiologic characteristics of an NB [12] promote an unfavorable respiratory condition in the process of ventilatory weaning, justifying the recommendation of TCH.

The major limitation of this study is the scarcity of studies on the pediatric indication of tracheostomy in neurologic children, a fact that emphasizes the importance of disseminating to the scientific scope the report of the experience encountered by health professionals in making a decision in a case of a child suffering from the Freeman-Sheldon syndrome.

## Disclosure

Research conducted in the Mandaqui Hospital, the partner

institution Universidade Nove de Julho.

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