

Anesthetic Management of an Infantile Patient with Unrecognized Hyaline Fibromatosis Syndrome

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Introduction

Hyaline fibromatosis syndrome (HFS) is a rare, autosomal-recessive disease, characterized by papular and nodular skin lesions, gingival hyperplasia, joint contractures and bone involvement to variable degrees. Variations in the *ANTXR2* gene leads to impaired extracellular matrix function and aberrant fibroblast activity leading to hyaline accumulation in tissues.⁽¹⁾ The disease has a relentlessly progressive course, with many patients exhibiting a severe failure to thrive from a young age.⁽²⁾ Due to temporomandibular and cervical joint contractures, stenosis of the nasal cavities and laryngopharynx, multiple nodules throughout the body including inside the airway, and gingival hyperplasia, anesthetic management of patients with HFS can be challenging.⁽³⁾ Understanding the implications of hyaline fibromatosis syndrome can lead to successfully securing and maintaining an airway in these high-risk patient populations.

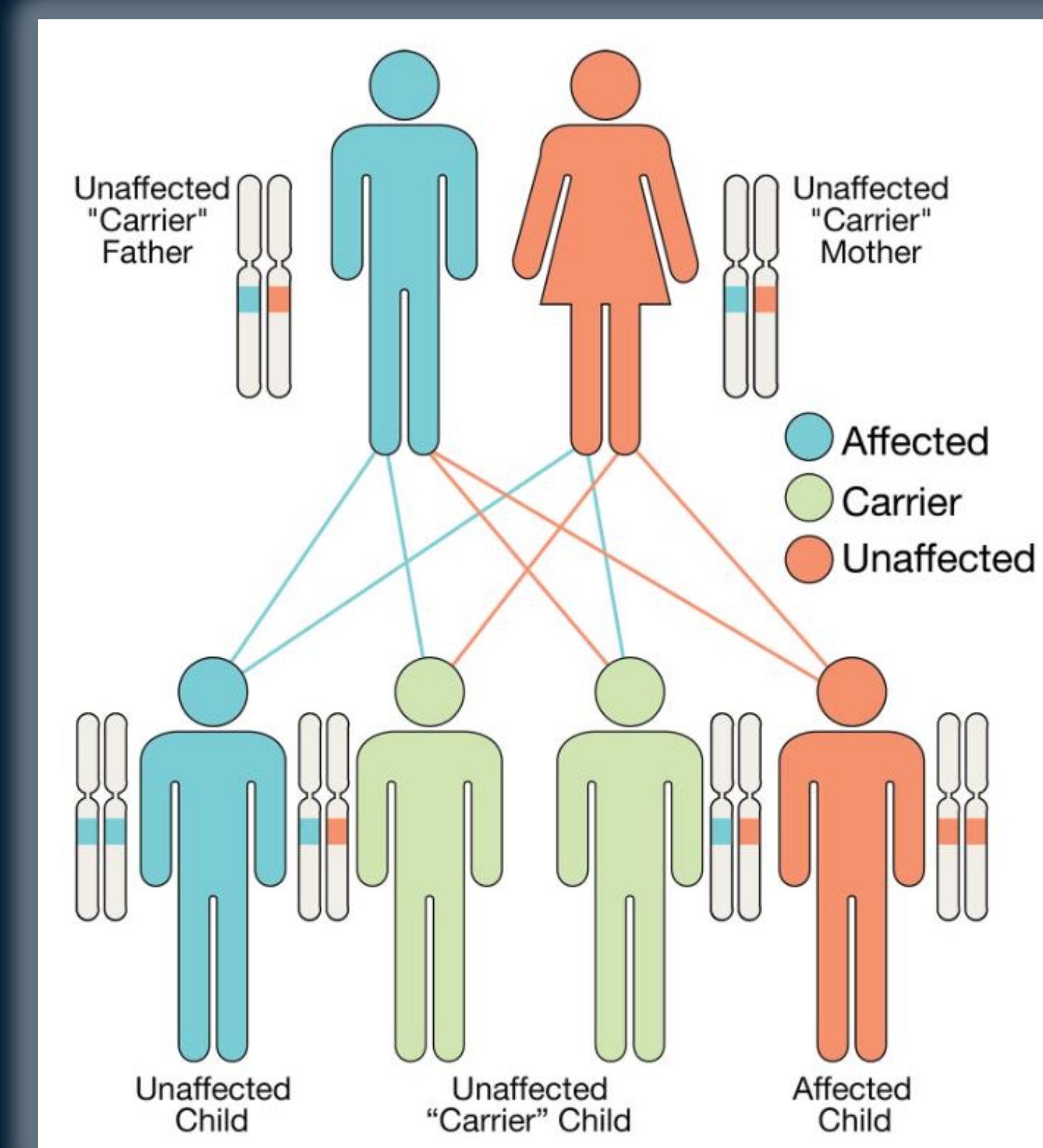


Figure 1: Depiction of the autosomal recessive pattern of inheritance of the *ANTXR2* gene.⁽⁴⁾

Case Report

Here, we present a six-month-old female with feeding intolerance, poor weight gain, and multiple contractures. Through genetic testing, it was discovered afterwards that she was born with unrecognized hyaline fibromatosis syndrome and clearly manifesting multiple sequelae: painful arthrogryposis, dysphagia, and failure to thrive. Prior to the diagnosis, a full body MRI was ordered to ascertain the involvement of the joint nodules and contractures.

We performed a rapid sequence induction with propofol and rocuronium due to the patient's persistent GERD. The first attempt to secure an airway was in MRI zone IV via direct laryngoscopy with an MRI-compatible Miller 1 blade. The best view obtained was a modified Cormack-Lehane grade 3. Intubation was not attempted, and we continued to easily mask ventilate and move the patient to zone III. The second attempt with a metal Miller 1 blade was met with the same result. Our third attempt was via video laryngoscopy with a C-MAC Miller 1 blade, providing a grade 2b view. We were able to successfully intubate, and placement was confirmed with auscultation and end-tidal monitoring. No adverse events such as airway trauma or aspiration occurred during the attempts. The patient was extubated and recovered appropriately in the post-anesthesia care unit.

Approximately one week later, the same patient was scheduled for a Nissen fundoplication and G-tube placement in the setting of severe GERD and nasogastric feeding dependence. The diagnosis of HFS was known at this time. The patient underwent rapid sequence induction with propofol and rocuronium. First attempt was with a C-MAC Miller 0 video laryngoscope. A grade 2b view was obtained. Initial insertion of the endotracheal tube was unsuccessful, but a quick adjustment of the curvature of the tube during easy mask ventilation allowed for successful and atraumatic passage. Placement was confirmed with auscultation and end tidal monitoring. The patient was also extubated successfully and recovered appropriately in the post-anesthesia care unit.

Discussion

Symptoms in patients with HFS can be variable and may increase in severity with age, making difficult airway management a significant anesthetic concern. Video laryngoscopy provided the best visualization of our patient's airway anatomy in both procedures. Yet, the best view obtained was a modified Cormack-Lehane grade 2b, displaying only the arytenoids. For our six-month old patient, no nodules were visualized around her airway, but the number of nodules can increase as patients with HFS age.⁽⁵⁾ Thus, caution should be applied for subsequent administrations of anesthesia. However, it is important to note that we were able to easily mask ventilate for both procedures albeit difficult airway anatomy for intubation. We recommend the use of video laryngoscopy, the presence of a highly experienced anesthesiologist, and prior imaging of the airway, if possible, for the anesthetic management of patients with HFS.

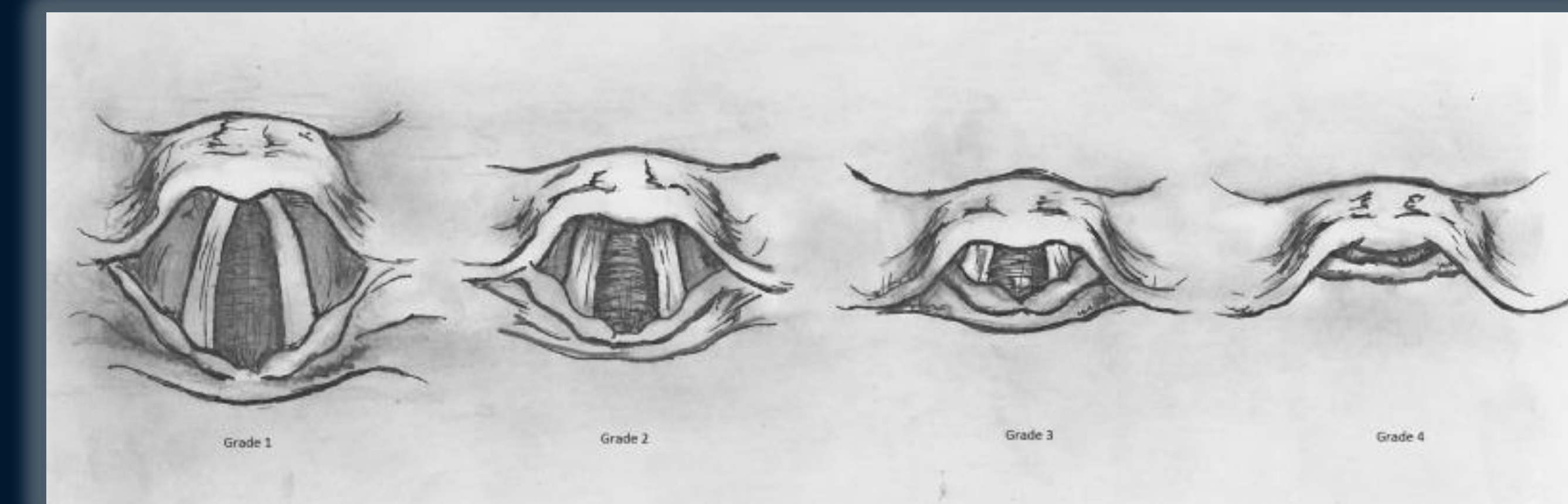


Figure 2: Cormack-Lehane Grading system as noted in Schrader et al: grade 1 - full view of glottis; grade 2 - partial view of glottis or arytenoids; grade 3 - only epiglottis is visible; grade 4 - no glottis structures are visible. In modified Cormack-Lehane (not pictured), grade 2 is split into 2a (partial view of glottis) and 2b (only arytenoids or very posterior origin of the cords are visible).⁽⁶⁾

References

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