Achondroplasia: Otolaryngologic Manifestations

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Overview

In this chapter, we provide an overview of otolaryngologic manifestations of achondroplasia. Particular attention should be given to upper airway obstruction, respiratory compromise and hearing loss.

Introduction

Achondroplasia is the most common genetic form of skeletal dysplasia, with an incidence ranging between 1:15,000 to 1:40,000 live births[1]. The disorder is inherited as an autosomal dominant trait, although at least 90% of the cases are sporadic and de novo mutations are associated with increased paternal age[2]. Achondroplasia is caused by an activating mutation in the transmembrane domain of fibroblast growth factor receptor 3 (FGFR3) where there is a single amino acid substitution, arginine for glycine, in amino acid 380[2,3]. The result is premature closure of growth plates and failure of endochondral ossification, leading to the skeletal characteristics of achondroplasia[4]: a long, narrow trunk with shortening of the proximal limbs (rhizomelia), large head with frontal bossing, and mid-face hypoplasia[5]. The term achondroplasia was proposed by Parrot in 1878, although earlier case reports have been noted. The diagnosis of achondroplasia is usually made by physical exam. Achondroplasia patients are at higher risk for respiratory and otologic difficulties.

Respiratory

Children with achondroplasia often have respiratory disorders. Respiratory concerns range from obstructive to central sleep apnea, as well as restrictive lung disease. Zucconi *et al* found that the most common cause of respiratory difficulties in achondroplasia children was due to upper airway obstruction. A short cranial base and midface hypoplasia as seen in achondroplasia play a more important role in respiratory difficulties than any other mechanism[6]. Onodera *et al* further clarified the etiology of upper airway obtstruction by attributing this to a retruded position of the chin, an increased mandibular plane angle, and an increased lower facial height due to an increased mandibular angle[7]. Due to the midface hypoplasia, there is "relative" tonsil and adenoid (T&A) hypertrophy which can further cause upper airway obstruction[8-9].

OSA

First-line management of OSA is generally with adenotonsillectomy. Adenoidectomy alone is generally not successful. [10] Success may be limited however with some children requiring positive airway pressure at night time via a CPAP or BiPAP apparatus.[11] Caution during the surgical procedure of T&A is warranted with respect to neck extension and smaller endotracheal tube size than generally seen for same-age children without Achondroplasia.[10] Preoperative assessment for foramen magnum stenosis is warranted.[12]

Central apnea may be caused by foramen magnum stenosis leading to cervicomedullary compression. Central apneas and sudden death in severe cases may result. Collins *et al.* found that 11 of their 22 tertiary care patients (50%) required decompression after being evaluated for foramen magnum stenosis[12].

Restrictive lung disease further complicates management for children with achondroplasia.[13] The small chest cage, characterized by short, flared ribs, causes respiratory complications[14,15]. Abnormal rib development reduces the lungs' elastic recoil and interferes with normal intercostal muscle function which may reduce the functional residual capacity (FRC). The reduced FRC along with reduced elastic recoil could cause airway closure, atelectasis, and hypoxemia.[15] In addition, the vital capacity (VC) of achondroplasia patients was only 67% of normal in Stokes *et al.*[11,15]

Otologic

Achondroplasia patients are at increased risk for otitis media and conductive hearing loss. Sensorineural and mixed hearing loss may also be seen.

Because of midface hypoplasia, the Eustachian tubes are short, the pharynx is small, and there is a relative hypertrophy of the adenoids causing impaired nasal airflow[16]. This predisposes children with achondroplasia to otitis media. Between 78-95% of children with achondroplasia in large studies suffered from OM. [16,17]

Bilateral myringotomy with tube insertion (BMT) is generally effective in the surgical management of OM in achondroplasia, with many such children undergoing this procedure. [16,18]

As with T&A, caution during surgery should be exercised to avoid excessive cervical spine hyperextension.

BMT usually corrects the conductive hearing loss (CHL) seen in achondroplasia.[8,19] Rarely, persistent CHL may be seen due to ossicular chain stiffness from congenital or chronic inflammatory etiologies. [20] here is disagreement on the prevalence of tAchondroplasia, and on the impact of these anomalies on hearing loss and chronic otitis media. [21,19,22,18]

Conclusion

Achondroplasia children require coordinated and proactive Otolaryngologic care. Airway and otologic manifestations of this disorder are generally successful with careful attention to the nuances required in caring for these patients. Awareness of cervical spine, tracheal and restrictive pulmonary disease allows for optimal surgical management.

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Additional recomended readings

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