Otolaryngologic Manifestations of Skeletal Dysplasias in Children

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KEYWORDS

• Skeletal dysplasia • Achondroplasia • Osteogenesis imperfecta • Hearing loss • Obstructive sleep apnea

KEY POINTS

- Children with skeletal dysplasias often present for otolaryngology evaluation; knowledge of the various syndromes and manifestations assists in diagnosis and management of ear, nose, and throat disease.
- Children with achondroplasia commonly have middle ear disease from eustachian tube dysfunction, and about half of adults and one-quarter of children with this skeletal dysplasia have hearing loss.
- Children with skeletal dysplasias are at high risk for development of obstructive sleep apnea. Although adenotonsillectomy is often the first-line treatment, the causes of sleep-disordered breathing in these patients are complex and the management is difficult.
- Children with skeletal dysplasias often undergo a variety of surgical procedures. They present unique anesthetic management issues because of variations in upper and lower airway anatomy, neck motion and stability issues, difficulties with chest and pulmonary mechanics, and abnormalities of neuromotor tone.

Skeletal dysplasias are a group of disorders of bone and cartilage development that result in abnormalities of the skeleton, with disproportionate growth of the long bones, cranium, and spine. With more than 350 types of skeletal dysplasias identified, skeletal dysplasias are estimated to affect 1 in 5000 births in the United States.[1] The most common nonlethal skeletal dysplasias are achondroplasia/hypochondroplasia, osteogenesis imperfecta (OI), variants of spondyloepiphyseal dysplasia, and

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pseudoachondroplasia; whereas thanatophoric dysplasia, severe OI, and achondrogenesis are common prenatal/perinatal lethal skeletal dysplasias.[2–4] Some skeletal dysplasias may be diagnosed before birth using high-resolution prenatal imaging in combination with prenatal molecular testing, but most are diagnosed at or after birth based on clinical and/or radiographic features and confirmed by molecular testing (**Table 1**).

Although skeletal dysplasias primarily manifest with short stature and orthopedic symptoms, not all are associated with short stature and many have associated otolaryngologic and even serious multisystem disease. These patients are best evaluated by a multidisciplinary team of specialists experienced in skeletal dysplasias, and head and neck manifestations of these disorders often require an otolaryngologist.

HEAD AND NECK MANIFESTATIONS

The skeletal dysplasia syndromes have a variety of head and neck manifestations. Otolaryngology evaluation of affected individuals can reveal obvious and less obvious ear, nose, and throat disease. The major issues of concern for most children with skeletal dysplasias involve the ears and hearing, and upper airway/respiratory function. An otolaryngologist experienced in the care of syndromic patients can provide specialized diagnostic services and surgical treatment of head and neck disorders associated with skeletal dysplasia. In some cases, otologic or upper airway respiratory symptoms may be the first presenting signs of these conditions.[5]

ACHONDROPLASIA

Achondroplasia is the most common syndrome of short-limb dwarfism. This autosomal dominant condition is caused by mutations in fibroblast growth factor receptor 3 (FGFR3).[6] Typical facial features include frontal bone prominence and midfacial hypoplasia, and short stature with rhizomelia is seen (**Fig. 1**). About 80% of children with achondroplasia are born to parents of average stature, indicating a new mutation in the affected child. Final height for adults with achondroplasia is slightly more than 1.2 m. Although motor delays are common in young children with achondroplasia because of macrocephaly in combination with axial and appendicular hypotonia, cognitive delays are not and require additional evaluation when identified. Almost 40% of children with achondroplasia had conductive hearing loss, and similarly 40% eventually underwent adenotonsillectomy, in one multicenter retrospective study of medical complications of achondroplasia.[7] Otolaryngologic manifestations of achondroplasia are addressed later in this article.

OTOLARYNGOLOGIC EVALUATION OF CHILDREN WITH SKELETAL DYSPLASIAS

Otolaryngologic symptoms associated with skeletal dysplasia may arise early in the neonatal period or may occur later in childhood. Otolaryngology care involves evaluation of symptomatic children with skeletal dysplasias as well as participation in routine anticipatory longitudinal medical care. The American Academy of Pediatrics Committee on Genetics has recommended, in a guideline published in 1995 and updated in 2005, that children with achondroplasia receive yearly hearing screens from infancy and undergo speech and language evaluation before 2 years of age.[8] This guideline also recommends routine screening for signs of sleep apnea, with a low threshold for testing with polysomnography and/or referral to sleep specialists. This committee suggested that such routine screening for hearing, speech, and sleep apnea is advisable in all other skeletal dysplasia diagnoses until proved to be unnecessary