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CASE REPORT

Achondroplasia: Craniofacial manifestations and considerations in dental management

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KEYWORD

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Abstract Achondroplasia is the most common form of skeletal dysplasia dwarfism that manifests with stunted stature and disproportionate limb shortening. Achondroplasia is of dental interest because of its characteristic craniofacial features which include relative macrocephaly, depressed nasal bridge and maxillary hypoplasia. Presence of large head, implanted shunt, airway obstruction and difficulty in head control require special precautions during dental management. Craniofacial manifestations and considerations in dental management are presented in 11-year-old female patient with achondroplasia.

1. Introduction

Achondroplasia is the most common form of short-limb dwarfism, occurring in 1 of 25,000 births (Orioli et al., 1986). It is inherited as an autosomal dominant trait; however, 80% of the cases are sporadic (Rousseau et al., 1994). Achondroplasia is caused by mutation in fibroblast growth factor 3 (FGFR3) on chromosome 4, causing a defect in the maturation of chondrocytes in the cartilage growth plate which enables abnormal cartilage growth-plate differentiation and insufficient bony development (Rousseau et al., 1994). Prenatal diagnosis of achondroplasia is usually suspected on routine ultrasound with the image of shortened long bones and can be confirmed by molecular testing (FGFR3 mutational testing) of prenatal specimens (Boulet et al., 2009). The diagnosis can usually be made on the basis of clinical characteristics and specific features on radiographs. Clinical features of achondroplasia include disproportionate short stature with normal trunk length, rhizomelic (the proximal end) shortening of the extremities, bowing of the lower extremities, short stubby trident hands (increased space between the third and the fourth fingers), spinal stenosis and lumbar lordosis (curved lower spine) (Shirley and Ain, 2009). Craniofacial characteristic of this disorder include macrocephaly, prominent forehead, depressed nasal bridge, maxillary hypoplasia, otolaryngeal system dysfunction, and foramen magnum stenosis (Shirley and Ain, 2009). These characteristics may lead to number of complications including hydrocephalus, apnea, upper-airway obstruction, otitis media, sinusitis and dental malocclusion (Steinbok et al.,

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1989; Hunter et al., 1998). Other complications may include obesity and diabetes (Hoover-Fong et al., 2008). There is no cure for this disease, however, extended limb lengthening has been used to improve stature (Aldegheri and Dall'Oca, 2001). Growth hormone therapy may result in a transient increase in growth rate but not effective in significantly increasing stature (Seino et al., 2000). Most individuals with achondroplasia are of normal intelligence and are able to lead independent and productive lives (Thompson et al., 1999).

There is no clear data about the prevalence of achondroplasia in Saudi Arabia and some Arab countries (Bittar, 1998), however, Al-Gazali et al. (2003) reported that the birth prevalence of achondroplasia in the United Arab Emirates was 1.05/10,000 which is higher than those reported for other populations (Orioli et al., 1986). The high prevalence of consanguinity in these countries may lead to an increase in the number of children with genetic disorders (El-Mouzan et al., 2008). It was predicted that children with genetic disorders including achondroplasia will seek dental care and consultation in general and specialized dental clinics. Dentists should be aware of the clinical characteristics of achondroplasia and the complications that may arise as a result of this genetic disorder.

The purpose of this case report is to present the systemic and the dentofacial features of achondroplasia and discuss special consideration that should be taken in the case of dental management of this condition.

2. Case report

An 11-year-old Saudi girl was referred from the genetic clinic at King Saud Medical Complex in Riyadh, Saudi Arabia to the Pediatric Dentistry Clinic. Her complaint was dental caries and bleeding gum. She is the eldest among 3 siblings. Parents were first degree cousins. The mother had full term pregnancy and was delivered by caesarian section. There was no history of similar family condition. Medical history revealed that the patient is a known case of achondroplasia. After birth, the patient was admitted to the Intensive Care Unit for one week because of suspected hydrocephaly. She was suffering from hypotonia and vitamin D deficiency. Vitamin D was prescribed for 18 months. The patient started to walk at 2 years of age. She complained of recurrent ear infection and tonsillitis. Adenoidectomy was performed when the patient was 7 years old. At age 10, she started complaining of hearing difficulty, back pain and numbness of feet. At present, she is under recall neurological visits. Dietary history was analyzed which include frequent daily sugar intake.

Figure 1 Achondroplastic girl with little stature and short limbs.

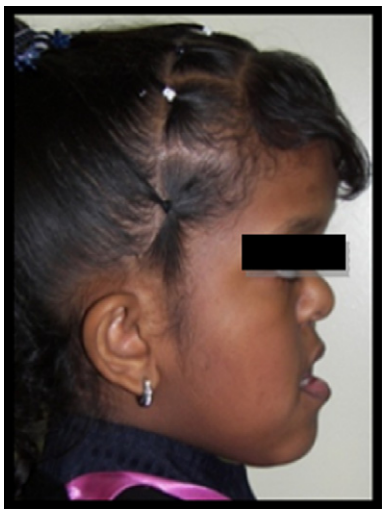

Figure 2 Lateral view showing concave profile and incompetent lip.

The patient appeared to be well adjusted, healthy and intelligent but had speech difficulty. She was 100 cm tall at the initial visit (Fig. 1). Extra-oral examination revealed classical manifestations associated with achondroplasia such as short stature, short stubby trident hands, frontal bossing, saddle nose, severe midface hypoplasia and incompetent lips (Figs. 2 and 3). Mouth breathing, interlabial gap of about 12 mm at rest and hyperactive mentalis were noticed. Lip closure was not possible without muscle strain. A concave facial pro-

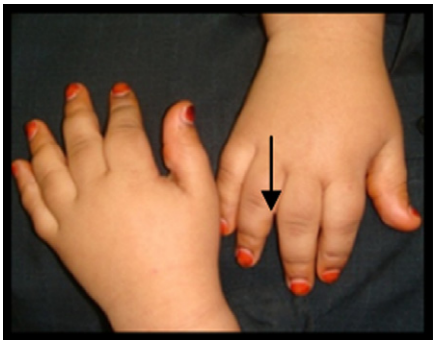

Figure 3 Short stubby trident hands (increased space between the third and the fourth fingers).

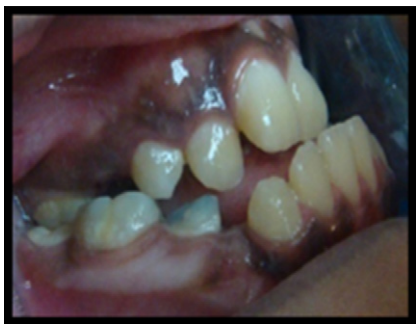

Figure 4 Intraoral views showing dental caries, anterior open bite, anterior reversed jet, posterior cross bite and class III dental malocclusion.

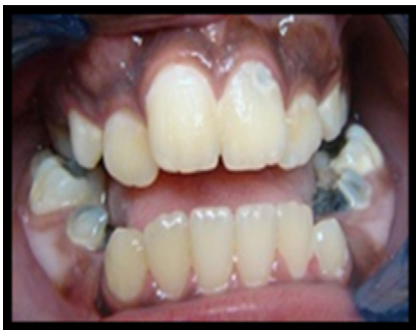

Figure 5 Intraoral views showing dental caries, anterior open bite, anterior reversed jet, posterior cross bite and class III dental malocclusion.

file was also noticed, however the mandible appeared normal and the chin was not prominent (Fig. 2).

Intraorally, macroglossia, tongue-thrust swallowing pattern, generalized gingivitis, posterior crossbite, anterior openbite and anterior reversed jet were observed (Figs. 4 and 5). She had dental class III molar relation with crowding at the maxillary anterior region. Patient had mixed dentition; the size, number and form of teeth were normal with multiple carious lesions and restorations. Eruption of teeth was compatible with the chronological age. Panoramic radiograph showed complete number of permanent teeth including third molars with normal development, crowding at the upper canine and molar regions. Cephalometric analysis of the lateral skull radiograph revealed that maxilla was severely retrognathic, with normal position of the mandible, decreased upper facial height, increased lower facial height and a skeletal class III jaw relation (Fig. 6). Dentally, the maxillary incisors were proclined and the mandibular incisors were normally inclined. Caries risk was assessed. Treatment plan was formulated to extract all carious primary teeth and to restore all carious permanent teeth (Figs. 7–9). Dental treatment was performed as planned under local anesthesia and the patient was cooperative. The patient was put under recall program every 3 months.

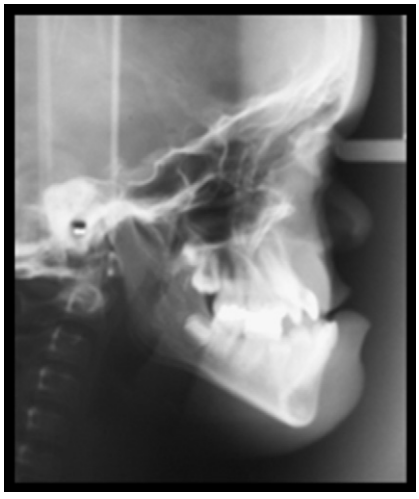

Figure 6 Lateral skull radiograph revealed retrognathic maxilla, with normal position of the mandible, and skeletal class III jaw relation.

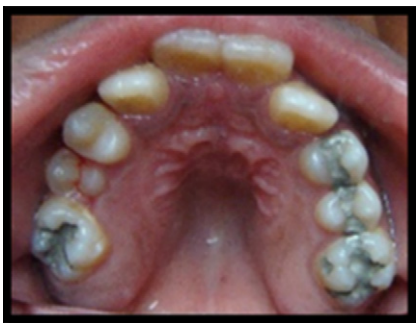

Figure 7 Post operative views after completion of dental treatment.

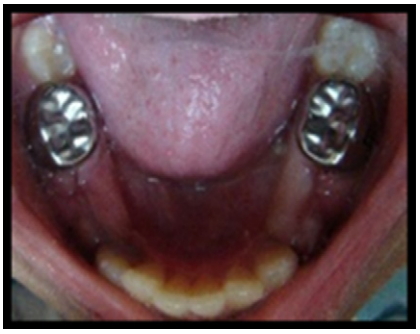

Figure 8 Post operative views after completion of dental treatment.

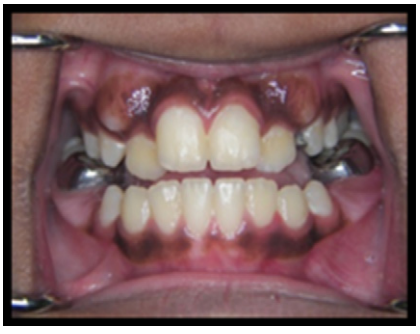

Figure 9 Post operative views after completion of dental treatment.

Orthodontic consultation was conducted revealing that it was too late to intercept orthodontically at this age, as the patient approaching puberty. Due to severe maxillary hypoplasia and skeletal class III, orthognatic surgery was decided to be performed after complete puberty. Initial orthodontic treatment plan was proposed to extract all first premolars. Le Fort III surgery for midface advancement was suggested to be performed after craniofacial team consultation. Fixed orthodontic appliance is to be placed after surgery.

3. Discussion

Although skeletal dysplasias are relatively rare, it is estimated that 250,000 individuals worldwide have achondroplasia, the most prevalent form of dwarfism (Horton et al., 2007). Other rhizomelic dwarfing disorders such as hypochondroplasia and thanatophoric dysplasia are part of the differential diagnosis. However, in comparison with achondroplasia the changes in hypochondroplasia are milder and the changes in thanatophoric dysplasia are much more severe and always lethal (Trotter and Hall, 2005).

Few studies were reported regarding the dentofacial findings, potential dental problems and its management in achondroplastic children (Ohba et al., 1998; Celenk et al., 2003; Stephen et al., 2005). The presented case showed the classical systemic manifestations and oral findings of achondroplasia.

This 11-year-old girl presented with a short stature (100 cm) which result from generalized defect in endochondral osteogenesis due to the deficiency of cartilage cell proliferation, particularly at the epiphyses of long bones which lead to short limbs. Early closure of suture at the skull base accounts for the small foramen magnum, short cranial base and J-shape sella turcica (Barone et al., 1994). Patient presented with severe midface hypoplasia causing psudoprognathism and depressed nasal bridge causing saddle nose deformity. These characteristics are believed to be the result of impaired endochondral bone formation and the membranous ossification proceeds normally (Rimoni et al., 1974). Because condylar cartilage is the product of periosteal chondrogenesis (Meikle, 1973), mandibular growth is not affected.

The patient started to walk at 2 years of age as children affected with achondroplasia commonly have mild to moderate hypotonia and development of motor skills is delayed (Trotter and Hall, 2005). She appeared to be intelligent as intelligence is almost certainly normal unless hydrocephalus or other central nervous system complications arise (Thompson et al., 1999). The patient was complaining of recurrent otitis media which results in gradual hearing loss in the left ear. The patient also had speech difficulty. The increased incidence of otitis media in achondroplasia due to short eustachian tubes necessitates annual hearing test and proper speech evaluation by 2 years of age (Hunter et al., 1998; Trotter and Hall, 2005; Collins and Choi, 2007). In this case, speech defect might have been resulted from articulating difficulty due to severe malocclusion.

Dental development can be delayed in achondroplastic children due to altered bone growth (Vaccaro and Albert, 2001). Onodera et al. (2005) reported no delay in the eruption of the primary and permanent teeth and it was according to the chronological age, which is consistent with this case. There was no available data regarding the incidence of dental caries in achondroplastic children. The patient in this case was at high risk for dental caries due to severe malocclusion, frequent intake of simple sugar and poor oral hygiene.

The patient manifested midface hypoplasia and constricted maxilla with relatively large mandible which result in skeletal and dental class III, posterior crossbite, anterior reversed jet and anterior open bite. According to Stephen et al. (2005), maxillary hypoplasia and relative mandibular prognathism were consistent features among achondroplastic children. Dunbar et al. (1980) concluded that the main orthodontic problem in achondroplasia is class III malocclusion. Early orthodontic evaluation should be considered in achondroplastic children to attempt the possibility of interceptive orthodontic, which was not applicable in the presented case. American Academy of Pediatrics recommends review of orthodontic problems in achondroplasia after 5 years of age (Trotter and Hall, 2005). Literature reported successful orthodontic treatment in achondroplastic children with or without orthognatic surgery (Dunbar et al., 1980; Barone et al., 1994; Ohba et al., 1998; Celenk et al., 2003; Stephen et al., 2005).

Dealing with achondroplastic children needs special psychological management during dental treatment, as the presence of disproportionate short stature can cause a number of psychosocial and social problems (Trotter and Hall, 2005). Presence of little stature and short limbs, in addition to chronic backache made it hard for this achondroplastic girl to sit comfortably on a conventional dental chair. Backache indicates spinal stenosis which is usually noticeable at late childhood and early adolescence (Hecht and Butler, 1990; Hunter et al., 1998). Lowering the dental chair and the use of step stool will help the achondroplastic child to get on the dental chair easily. A cushion behind the child's back may be required during dental treatment for good posture and to reduce back pain.

Although there was no shunt implanted in the presented case, shunt was reported to be placed in some achondroplastic children due to hydrocephalus (Hunter et al., 1998; Stephen et al., 2005) which makes head control difficult and may necessitate antibiotic prophylaxis before dental treatment (Tong and Rothwell, 2000). Furthermore, special precautions in head control during dental intervention are essential, due to the possible presence of cranio-cervical instability, foramen magnum stenosis and limited neck extension, as it might result in respiratory complications. This is very important especially when dealing with an uncooperative achondroplastic child. In addition, large tongue and relatively large mandible may lead to the increased difficulty of airway management in case of emergency. This patient was also complaining of enlarged tonsils and adenoids causing upper-airway obstruction which resulted in mouth breathing, and subsequently generalized gingivitis (Wagaiyu and Ashley, 1991). Mouth breathing due to upper-airway obstruction was reported to be a constant feature in achondroplastic children (Onodera et al., 2005; Stephen et al., 2005).

As in this case, it is advised to perform dental treatment under local anesthesia, because general anesthesia poses certain complications due to anteriorly placed epiglottis, small nasal pharynx and larynx, difficulty in intubation, lumbar lordosis, narrowing of spinal cord and small chest (Kalla et al., 1986; Butler et al., 2000). It is important for dental professionals, including pediatric dentists, orthodontists and oral surgeons treating these patients, to recognize risk factors and potential complications before sedation or anesthesia. When dental treatment is decided to be under GA, it is recommended to

do radiologic evaluation of foramen magnum, preoxygenation before anesthesia, using appropriate endotracheal tube size, oral intubation and administration of oxygen after extubation (Kalla et al., 1986).

In conclusion, the presented case showed that the features of achondroplasia may lead to respiratory, neurological, skeletal, orthodontic and psychosocial problems. Dentists treating these children should be able to recognize these features and its complications as dental management is constrained by practical problems associated with this disease.

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