ARTICLE

ABSTRACT

Goldenhar's syndrome is a rare disorder characterized by several anomalies that include dermal epibulbar cysts, auricular appendices and malformations and vertebral anomalies. In this article, the authors report a case of Goldenhar's syndrome in a 10-year-old child who presented with the classical signs of this condition and a solitary median maxillary central incisor (SMMCI).

KEY WORDS: Goldenhar's syndrome, dental treatment, solitary median maxillary central incisor

Solitary median maxillary central incisor in association with Goldenhar's syndrome: a case report

Francisco Wanderley Garcia de Paula e Silva, DDS;[1] Fabrício Kitazono de Carvalho, DDS;[1] Kranya Victória Díaz-Serrano, DDS, MSc, PhD;[2] Aldevina Campos de Freitas, DDS, MSc, PhD;[2] Maria Cristina Borsatto, DDS, MSc, PhD;[2] Alexandra Mussolino de Queiroz, DDS, MSc[2*]

[1]Postgraduate Student; [2]Professor — Department of Pediatric Clinics, Preventive and Social Dentistry, Faculty of Dentistry of Ribeirão Preto, University of São Paulo, Brazil. *Corresponding author e-mail: amqueiroz@forp.usp.br

Spec Care Dentist 27(3): 105-107, 2007

Introduction

Goldenhar's syndrome is a rare disorder characterized by numerous anomalies affecting the first and second branchial arches.[1] The incidence of this condition, also known as oculoauriculovertebral dysplasia or hemifacial microsomia,[2] is about 1 in 5,000 to 25,000 live births. This syndrome occurs at a higher rate in males than females.[3]

The etiopathology of Goldenhar's syndrome is not clear. Possible causes of this disorder range from disturbance of the neural crest cells during embryogenesis[4] to chromosomal abnormalities.[5] Systemic and environmental conditions during pregnancy, i.e. maternal diabetes and use of drugs such as cocaine, thalidomide and retinoic acid, have also been associated with the development of this syndrome.[6,7]

Commonly observed malformations of Goldenhar's syndrome include ocular and auricular anomalies, such as microtia, anotia, dermal epibulbar tumors, vertebral anomalies, unilateral maxillary and malar hypoplasia, palatal clefts, and central nervous system disturbances.[6,8]

Developmental disturbances during initiation and proliferation stages of tooth formation are known to produce aberrations in both the number and the structure of teeth. In the permanent dentition, hypodontia (congenital absence of teeth) is most commonly seen in third molars, followed by second premolars and maxillary lateral incisors. The occurrence of a single maxillary permanent central incisor in the midline is a rare developmental anomaly. The frequency of a solitary median maxillary central incisor (SMMCI) has been estimated as 1:50,000.[9]

Although the exact mechanism responsible for the occurrence of SMMCI is still unknown, it has been suggested[10] that agenesis of the contralateral tooth germ may cause the eruption of a single central incisor in the midline. Other theories have proposed[11] that the formation of one central incisor instead of two could result from disturbances in the mitotic potential of the incisor group, which would be influenced by genetic or environmental factors. It has also been suggested that space limitation within the dental arch or deficiency of lateral growth from the midline would result in premature fusion of the spreading dental lamina epithelium from left and right sides in the maxilla. As a consequence, this would prevent the formation of the mesial halves of the central incisors and result in a single tooth consisting of the fused distal halves of the permanent left and right central incisors.[9]

The occurrence of a SMMCI was first

described in a 6-year-old child.[12] Since then, it has been reported[10,13] either as an isolated finding or associated with a variety of midline developmental defects, holoprosencephaly and/or growth deficiency.

This article reports the case history of a boy with Goldenhar's syndrome who presented with the classical signs of the syndrome and a solitary median maxillary central incisor.

Case report

A 10-year-old male patient with Goldenhar's syndrome was brought for general dental care to the clinic of the Center of Formation of Human Resources Specialized in Dental Care for Special Patients, at the Faculty of Dentistry of Ribeirão Preto, University of São Paulo, Brazil.

The patient was born to healthy nonconsanguineous parents after a full-term, uneventful pregnancy. The medical history was noncontributory and the mother denied using drugs during pregnancy. There was no history of orofacial trauma at any time. The only intervention found in the medical history in the region of the head was a surgical procedure eight days after birth for placement of an intracranial valve due to congenital hydrocephaly.

An extraoral examination revealed facial asymmetry, indistinct philtrum, auricular anomalies and absence of the left eyeball (Figure 1). An intraoral examination revealed the presence of a solitary median maxillary central incisor, absence of upper lip frenulum (Figure 2) and incisive papilla (Figure 3). In addition, the patient had teeth with extensive carious lesions that needed immediate restorative treatment.

An evaluation of the panoramic radiograph confirmed the existence of a single maxillary central incisor and also showed the presence of impacted teeth (#13 and 29), the existence of a hypoplastic left coronoid process of the mandible, and the absence of other dental anomalies (Figure 4).

The dental treatment plan was based on the patient's high caries risk and consisted of extraction of teeth #3 and 19, restoration of teeth #14 and 18 with amalgam and teeth #10 and 30 with composite resin. As part of the treatment strategy for a high caries risk patient, resin-based sealants were placed on teeth #4 and 5.

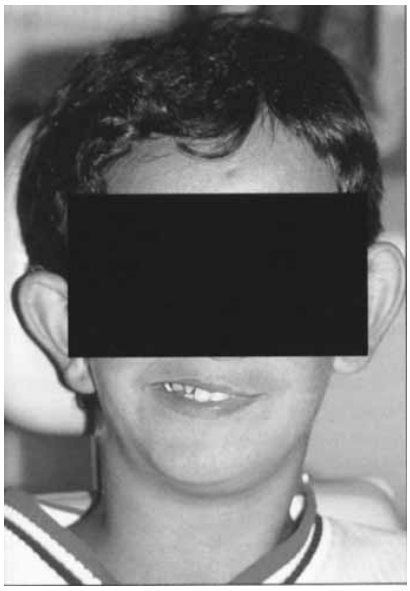

Figure 1. Frontal view of the patient showing facial asymmetry, indistinct philtrum and absence of the left eyeball.

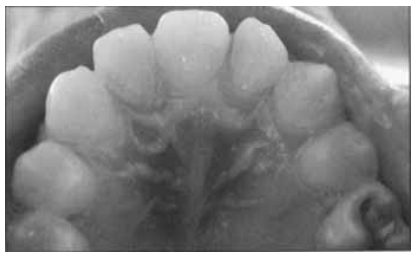

Figure 3. Intraoral occlusal view showing the single maxillary central incisor and absence of incisive papilla.

Although neither the patient nor his parents voiced any concern about the esthetics of the SMMCI, the restorative possibilities were presented to them and discussed. The parents were instructed that, if the patient had esthetic concerns at a later age, the alignment and shape of the anterior teeth could be altered to improve symmetry and harmony. However, successful care would depend on careful joint orthodontic and cosmetic planning.

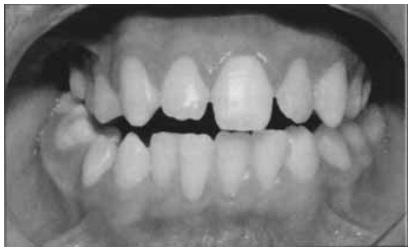

Figure 2. Intraoral frontal view of the solitary median maxillary permanent central incisor showing the absence of upper lip frenulum.

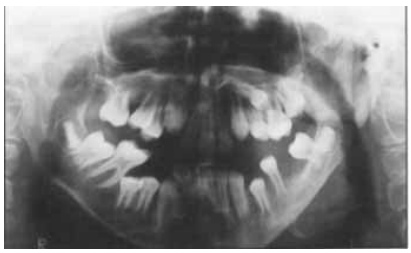

Figure 4. Panoramic radiograph confirming the existence of a single maxillary central incisor and also showing the presence of impacted teeth (#13 and 29) and a hypoplastic left coronoid process of the mandible. No other dental anomalies were observed.

Discussion

Patients with Goldenhar's syndrome may have a large number of malformations. The child presented in this case report had facial asymmetry and a hypoplastic mandible, which are typical characteristics of this disorder.[4,8] Although the frequency of cardiac disturbances in individuals with Goldenhar's syndrome ranges from 5% to 58%,[14] no cardiac problems were found in our patient.

Because there was no esthetic complaint, no restorative procedures were carried out to modify the form of the teeth adjacent to the SMMCI. Extraction of the impacted teeth (#13 and 29) also was not planned because they were asymptomatic and no visible pathological changes were seen that required the surgical removal of the teeth. We agree with Hicks,[15] who does not recommend the prophylactic extraction of impacted teeth and advised more conservative management based on a watchful waiting approach with regular recalls and radiographic follow-up.

Tooth agenesis may be an isolated finding or a clinical manifestation of a number of syndromes, some of which have specific patterns of hypodontia.[16] Most studies have associated the occurrence of a solitary median maxillary central incisor with growth hormone deficiency,[10,13] but there is no scientifically based relation between Goldenhar's syndrome and the eruption of a single maxillary central incisor in the midline. While children with SMMCI usually have short stature and low growth hormone levels,[9] the patient described in this report had normal height and development.

In our patient, congenital agenesis of one of the maxillary central incisors led the remaining central incisor to erupt in the midline, as has been previously reported by Yassin and El-Tal.[10] Due to the presence of a median single central incisor, the child did not have the upper lip frenulum and incisive papilla and had an indistinct philtrum, which are characteristics consistent with the findings by Kjaer *et al.*[17]

The exact mechanism responsible for this dental anomaly remains unclear. However, in view of the size and shape of our patient's SMMCI, it may be speculated that it originated from the fusion of the dental lamina epithelium, forming a tooth that consisted of two fused distal halves.[9]

The SMMCI is also considered a risk indicator for holoprosencephaly or might be a moderate manifestation of the dominant form of this condition.[18,19] Therefore, it is advisable that patients presenting with a solitary median maxillary central incisor undergo a detailed medical history and, if necessary, are referred for medical examination, in order to evaluate whether the single central incisor is an isolated trait or has an association with any syndromic or systemic condition.

References

1. Scholtz AW, Fish JH III, Kammen-Jolly K, *et al.* Goldenhar's syndrome: Congenital hearing deficit of conductive or sensorineural origin? Temporal bone histopathologic study. *Otol Neurotol* 2001;22:501-5.
2. Pinheiro AL, Araújo LC, Oliveira SB, Sampaio MCC, Freitas AC. Goldenhar's syndrome — case report. *Braz Dent J* 2003;14:67-70.
3. Rollnick BR, Kaye CI, Nagatoshi K, Hauck W, Martin AO. Oculoauriculovertebral dysplasia and variants: phenotypic characteristics of 294 patients. *Am J Med Genet* 1987;26:361-75.
4. Rodríguez JI, Palacios J, Lapunzina P. Severe axial anomalies in the oculo-auriculo-vertebral (Goldenhar) complex. *Am J Med Genet* 1993;47:69-74.
5. Wilson GN, Barr M Jr. Trisomy 9 mosaicism: Another etiology for the manifestations of Goldenhar Syndrome. *J Craniofac Genet Dev Biol* 1983;3:313-6.
6. Lessick M, Vasa R, Israel J. Severe manifestations of oculoauriculovertebral spectrum in a cocaine exposed infant. *J Med Genet* 1991;28:803-4.
7. Wang R, Martinez-Frias ML, Graham JM Jr. Infants of diabetic mothers are at increased risk for the oculo-auriculo-vertebral sequence: a case-based and case-control approach. *J Pediatr* 2002;141:611-7.
8. Altamar Rios J. Síndrome de Goldenhar: a propósito de um caso. *An Otorrinolaringol Iber Am* 1998;25:491-7.
9. Hall RK, Bankier A, Aldred MJ, Kan K, Lucas JO, Perks AG. Solitary median maxillary central incisor, short stature, choanal atresia/midnasal stenosis (SMMCI) syndrome. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 1997;84:651-62.
10. Yassin OM, El-Tal YM. Solitary maxillary central incisor in the midline associated with systemic disorders. *Oral Surg Oral Med Oral Pathol Radiol Endod* 1998;85:548-51.
11. Osborn JW, Ten Cate AR. *Advanced dental histology.* 4th ed. Boston: Wright/PSG; 1983:35-45.
12. Scott DC. Absence of upper central incisor. *Br Dent J* 1958;104:247-8.
13. Fleming P, Nelson J, Gorlin RJ. Single maxillary central incisor in association with mid-line anomalies. *Br Dent J* 1990;168:476-9.
14. Nakajima H, Goto G, Nakata N, Ashiya H, Ibukiyama C. Goldenhar syndrome associated with various cardiovascular malformations. *Jpn Circ J* 1998;62:617-20.
15. Hicks EP. Third molar management: a case against routine removal in adolescent and young adult orthodontic patients. *J Oral Maxillofac Surg* 1999;57:831-6.
16. Jorgenson RJ. Clinician's view of hypodontia. *J Am Dent Assoc* 1980;101:283-6.
17. Kjaer I, Becktor KB, Lisson J, Gormsen C, Russell BG. Face, palate and craniofacial morphology in patients with a solitary median maxillary central incisor. *Eur J Orthod* 2001;23:63-73.
18. Muenke M, Gurrieri F, Bay C, *et al.* Linkage of a human brain malformation, familial holoprosencephaly, to chromosome 7 and evidence for genetic heterogeneity. *Proc Natl Acad Sci USA* 1994;91:8102–6.
19. Odent S, Attie-Bitach T, Blayau M, *et al.* Expression of the Sonic hedgehog (SHH) gene during early human development and phenotypic expression of new mutations causing holoprosencephaly. *Hum Mol Genet* 1999;8:1683–9.