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Osteoglophonic dysplasia: dental and orthodontic implications

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Structured Abstract

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Aims and Objectives – Documentation of dental and orthodontic implications of osteoglophonic dysplasia (OGD).

Settings and Participants – Case report describing oral and dental manifestations of a female with OGD, aged 39 years, who was first documented three decades ago.

Results – This rare genetic disorder manifests with gross stunting of stature, associated with severe craniofacial malformation and multiple unerupted teeth. Radiographically, multiple lucent lesions were present in the tubular bones and mandible as well as several impacted teeth.

Conclusion – We concluded that prosthetic dental replacement in this patient would be difficult because of the distorted jaw relationship and large alveolar ridges. Equally, craniofacial reconstruction might be compromised by obstruction of the nasal airways, difficulty in intubation and postoperative respiratory problems.

Key words: case report; craniofacial abnormalities; diagnosis; growth disorder; impacted; jaw abnormalities; oral; tooth

Introduction

Osteoglophonic dysplasia (OGD) is a rare heritable skeletal disorder in which gross stunting of stature is associated with severe craniofacial abnormalities. The term 'osteoglophonic' pertains to the radiological appearance of multiple lucent regions in the skeleton (1). Failure of eruption of the teeth and mandibular prognathism are prominent oral manifestations. These components of the disorder, together with possible implications for general anaesthesia, are of importance in dental and orthodontic management. Hypertrophy of the gums may occur, and it has been

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suggested that giant cell granulomata may be involved (2).

An affected girl was initially seen in Cape Town in 1976, and in 2005 we had the opportunity to reassess her clinical and dental status. This latter aspect of OGD has not previously been discussed in the dental literature. Our findings are presented in this case report, together with a review of the literature and suggestions for orthodontic management.

Patient report

The affected female was born in Cape Town into a family of mixed ancestry in 1966. Craniofacial abnormalities were evident at birth, feeding was difficult during infancy and walking only commenced at the age of 3 years. She was examined in 1976 at the former Princess Alice Orthopaedic Hospital. At this time she had severe short stature (height 89 cm) and rhizomelic limb shortening. The major craniofacial manifestations were mandibular prognathism, bossing of the forehead (Fig. 1), relative midfacial hypoplasia, hypertelorism and wide everted nostrils (Fig. 2). Her palate was high and the teeth had remained unerupted. At the initial examination, her eyes and intelligence were normal, as were her central nervous system and cardiovascular system.

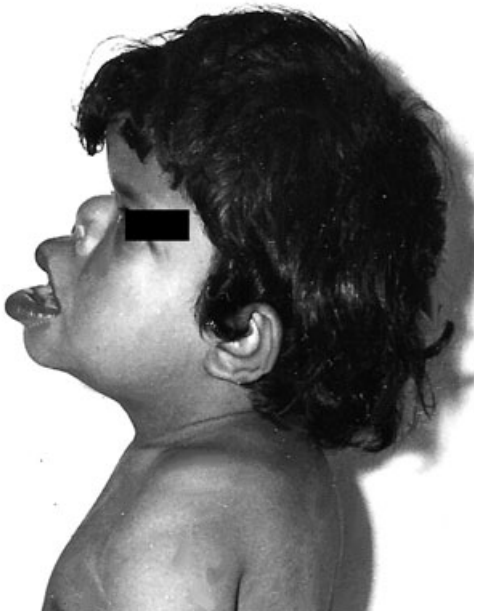

Fig. 1. The affected girl in 1976 at age 10 years. Gross mandibular hyperplasia and prominence of the forehead are evident.

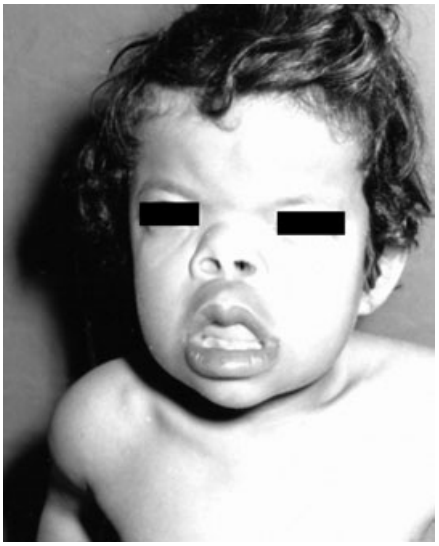

Fig. 2. The mouth and nostrils are wide and hypertelorism is present.

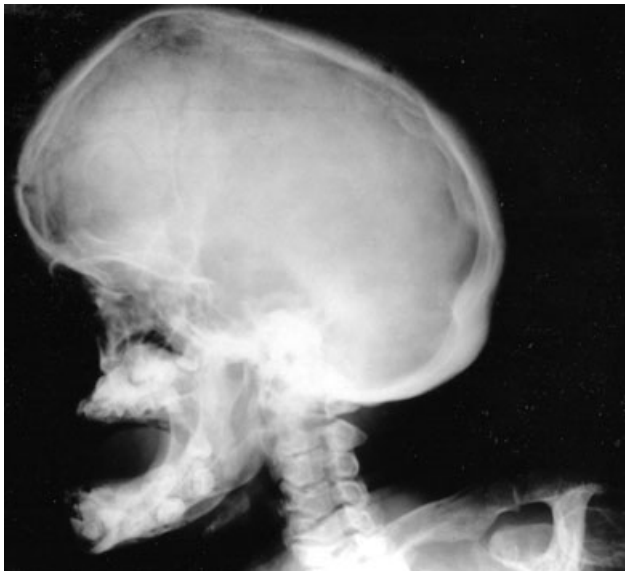

Fig. 3. Lateral skull radiograph showing multiple impacted teeth, some of which are inverted.

Radiographically the cranium was elongated, with frontal bossing. The ramus of the mandible was porotic with lucent cystic regions and many unerupted teeth were present (Fig. 3). The vertebral bodies were flattened and the pelvis was distorted. The tubular bones lacked the normal configuration, with multiple irregular lucent regions, maximal in the metaphyses. Her manifestations were subsequently documented and depicted in the radiological literature (1), and in a medical textbook (3).

Her father was 39 years of age at the time of her birth and the mother was aged 35 years. The family history was otherwise unremarkable: her three brothers and a sister were unaffected, as were her non-consanguineous parents and other family members.

When re-examined in 1988 and 2005, her appearance was essentially unchanged. She remained in good health, apart from difficulty in walking because of painful knee joints. She lived with a married sibling and occupied herself with needlework.

Dental observations

The patient was edentulous and tooth eruption had never occurred. Both maxillary and mandibular alveolar ridges were firm and fibrotic with gingival hyperplasia in the anterior mandibular area. She had a large tongue and a high palatal vault, but otherwise there was no obvious intra-oral pathology. On close examination of the skull radiographs, it was evident that a large proportion of the unerupted teeth were inverted and the roots poorly formed (Fig. 3).

Dental treatment implications

The most common dental manifestation in OGD is the absence of teeth, which results in functional difficulty and poor aesthetics. Treatment options include the construction of prostheses. The success of an intervention of this nature will depend on the structural relationship of the maxilla to the mandible and the amount of intra-oral space available for a prosthesis; in particular, the large alveolar ridges may reduce the amount of available space, while the presence of a large tongue may compromise the stability of a lower denture. Corrective surgery for craniofacial deformities warrants consideration, but the short stature and possible airway obstruction as a result of choanal atresia may compromise intubation and anaesthesia. Some affected persons have a predisposition to pneumonia, possibly related to disturbed respiratory function and atelectasis. The risks of this complication have implications for postoperative management.

Discussion

Osteoglophonic dysplasia was delineated by Beighton et al. (1) and earlier sporadic cases were subsequently recognized in the literature (4,5). Thereafter, Kelley et al. (6) documented an affected father and son, and Santos et al. (7) reported an affected girl. The condition was reviewed by Beighton (8) and additional cases were reported by Sklower Brooks et al. (9) and Azouz and Kozlowski (10). White et al. (2) elucidated the molecular defect and added four additional cases, including a father and son with the disorder, bringing the total of affected persons who had been reported to 12.

Clinical details provided in published case reports are not always comprehensive, and for this reason it is not possible to undertake objective numerical analysis of syndromic components. Nevertheless, it is evident that the major clinical manifestations are extreme stunting of stature, gross mandibular prognathism and pronounced frontal bossing. Hypertelorism, proptosis, a wide nose and relative midfacial hypoplasia are other significant craniofacial abnormalities. In the oral cavity, absence of tooth eruption is an important syndromic component while gingival hypertrophy, a high palate and macroglossia are variable features. Nasal obstruction is frequent and choanal atresia has been documented. Additional manifestations which have been reported include dysfunctional swallowing, nasal discharge and chronic gingivitis. Radiographically, the skeleton is dysplastic, with cystic lucent lesions predominantly situated in the metaphyses of the tubular bones and jaws.

The natural history of OGD is variable; death in infancy took place in a Portuguese girl (7), while at the other end of the spectrum, the South African patient whom we studied enjoyed good general health at the age of 40 years. Potentially lethal respiratory complications have been documented (2), but it is uncertain as to whether or not all affected persons are at risk in this way.

Failure of eruption of the teeth is a consistent manifestation of OGD. It is relevant, however, that although unerupted teeth are a component of more than 20 separate syndromes, OGD has not previously been mentioned amongst these disorders. Disturbances in tooth eruption are commonly the result of mechanical interferences caused by supernumerary teeth, crowding, and soft-tissue impaction as well as

by odontogenic tumours and cysts (11). The aetiology of unerupted teeth occurring in individuals affected with OGD remains speculative; the phenomenon could arise from or be complicated by the presence of cystic lesions of the jaw or the presence of inverted teeth.

Biopsy of a swelling in the lower gums of an affected individual revealed the histological features which were consistent with giant cell granuloma (2); it is possible that the cystic lesions in the jaw and long bones represent a similar process. The situation is complex, however, as other jaw lesions with similar radiographic features include odontogenic keratocysts, ameloblastoma, dentigerous cysts, aneurismal bone cysts and cherubism. It is evident that giant cell lesions represent a spectrum of disorders varying in their aetiology from reparative processes to manifestations of systemic disorders. The brown tumour of hyperparathyroidism represents a local manifestation of a systemic metabolic disease that is histologically and radiographically indistinguishable from a giant cell tumour (12). In this context, it may be relevant that there is some evidence for involvement of phosphate and vitamin D metabolism in OGD, and that the cystic lucent lesions in the skeleton are consistent with abnormalities in this biochemical pathway.

Osteoglophonic dysplasia is inherited as an autosomal dominant trait. The condition results from mutation in the fibroblast growth factor receptor 1 gene, and there is intragenic heterogeneity. In this context the specific molecular defect has been elucidated (13). Because of the severity of the manifestations, fitness to reproduce is diminished and the majority of affected individuals are sporadic, representing new mutations of the determinant gene. Nevertheless, two instances of affected fathers producing affected offspring have been reported (2,6). Currently, there is insufficient data for genotype–phenotype correlations, but it may eventually be possible to infer potential severity and consequent orthodontic prognosis from identification of the specific determinant mutation.

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