

CASE REPORT

Nonsyndromic Familial Hypodontia in Four Members of a Family: A Case Report

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ABSTRACT

Hypodontia constitutes one of the most common developmental anomalies in humans. Depending upon its severity, it can have a great impact on the oral health of the affected individuals. The present report concerns four members of a family with hypodontia. Whereas three members had unilateral missing maxillary lateral incisor only, the fourth member had bilateral missing maxillary lateral incisors and right mandibular second premolar. Their general examination did not suggest any syndrome present. On the basis of familial incidence and the absence of any clinical features associated with any syndrome, the final diagnosis of nonsyndromic familial hypodontia was made.

Keywords: Congenital absence, Familial, Hypodontia, Nonsyndromic, Maxillary lateral incisor.

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INTRODUCTION

The congenital absence of teeth is one of the commonest developmental abnormalities seen in human populations. Different terms are used to define the congenital absence of teeth. The developmental absence of one to six teeth (excluding third molars) is termed as hypodontia. Oligodontia refers to the absence of more than six teeth (excluding third molars) whereas anodontia refers to the complete absence of teeth.¹⁻³ The prevalence of hypodontia in the permanent dentition (excluding third molars) has been reported to range from 2.3–9.6%.⁴ Hypodontia can either occur in association with various genetic syndromes like Down syndrome, ectodermal dysplasia, Ehlers–Danlos syndrome, Turner syndrome,

or as an isolated nonsyndromic condition which can occur either sporadically or in a familial fashion.^{5,6} Although there have been reports of missing teeth in family members but most of these studies have reported cases with four to six missing teeth or oligodontia.⁷⁻¹³ The present article reports a case of hypodontia involving permanent teeth in four members of a family. The unique feature of this report is that three members of a family consistently had single maxillary lateral incisor missing with concomitant microdontia of contralateral maxillary lateral incisor. However, in the fourth member three teeth were found to be congenitally missing.

CASE REPORT

A 15-year-old boy reported to the Department of Dentistry with a chief complaint of spacing in between upper front teeth. When questioned about similar presentation in the family, it was learned that there was a history of missing teeth and spacing in other members of the family also. The patient had two elder sisters. Both the sisters as well as his mother had missing teeth as informed by the patient. Since the patient belonged to the family of medical professionals, he was well aware of this condition. The patient's past medical and dental history was not significant. The general examination did not suggest any syndrome present. On intraoral examination, the patient had missing right maxillary lateral incisor with microdontia of the maxillary left lateral incisor (Fig. 1). An orthopantomogram (OPG) was taken which showed the congenital absence of right maxillary lateral incisor with the presence of all third molars. (Fig. 2). Comprehensive orthodontic treatment was advised to the patient. The patient was asked to bring his mother and two sisters during his next visit, to confirm the familial pattern.

Mother: Patient's mother aged 51 years gave a history of surgical removal of right maxillary canine at 16 years of age as it was embedded and removal of upper third molars due to caries, at the age of 35. Further history of the patient revealed that her father also had some spacing and missing teeth. The intraoral examination showed missing left maxillary lateral incisor with left maxillary central incisor and canine in proximal contact with each other and microdontia of right maxillary lateral incisor. Besides it also showed missing right maxillary canine

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Figs 1A to C: Intraoral photographs of the patient



Fig. 2: OPG of a patient showing missing 12

with its extraction space present (Fig. 3). Suspecting congenital absence of left maxillary lateral incisor, an OPG was taken, which confirmed the absence of left maxillary lateral incisor with the presence of mandibular third molars (Fig. 4).

Sister 1: Patient's eldest sister aged 27 years gave the history of previous orthodontic treatment seven years back but revealed that no extraction was carried out during the treatment. Intraoral examination showed missing left maxillary lateral incisor with left maxillary central incisor and canine in proximal contact with each other and microdontia of right maxillary lateral incisor (Fig. 5). On our request from the orthodontist who treated her, we were able to get a pretreatment OPG. Unfortunately, pretreatment intraoral photographs could not be retrieved. The current OPG showed the absence of left maxillary lateral incisor with the presence of all the other teeth including third molars (Fig. 6A). The preorthodontic treatment OPG also showed missing left maxillary lateral incisor and microdontia of right maxillary lateral incisor (Fig. 6B). Hence this finding was confirmed.

Sister 2: Patient's second sister was undergoing fixed orthodontic treatment at the time of presentation, so she was advised to bring all the pretreatment records from

the treating orthodontist. The pretreatment intraoral photograph taken at the age of 13 revealed bilaterally retained deciduous maxillary second molar, deciduous left maxillary canine, and deciduous left mandibular second molar and extraction space of deciduous right mandibular second molar and unerupted both maxillary lateral incisors (Fig. 7). OPG showed congenitally missing maxillary lateral incisors bilaterally and right mandibular second premolar (Fig. 8). All third molars were present in various stages of development.

DISCUSSION

Nonsyndromic hypodontia is by far the most common form of congenital tooth absence and can involve a variable number of teeth.⁶ Although it can be caused by environmental factors and/or genetic factors, in the majority of cases hypodontia has a genetic basis. Mutation of one or more genes can cause hypodontia and often results in the prevalence of hypodontia among several members of the same family in multiple generations.¹⁴ Familial hypodontia is most often transmitted in an autosomal dominant pattern with incomplete penetrance and variable expressivity.^{15,16} Besides this, other modes of transmission like autosomal recessive, X-linked dominant or recessive patterns and polygenic or multifactorial inheritance patterns have also been proposed.^{6,16} In the present case, since all the three children and their mother, exhibited at least a single missing tooth and none showed any significant finding related to any syndrome, so the present case was diagnosed as nonsyndromic familial hypodontia. Patient's father did not have any missing tooth and patient's maternal grandfather had a spaced dentition and some missing teeth as informed by the patient's mother. This suggested that the affecting gene probably



Figs 3A to C: Intraoral photographs of patient's mother

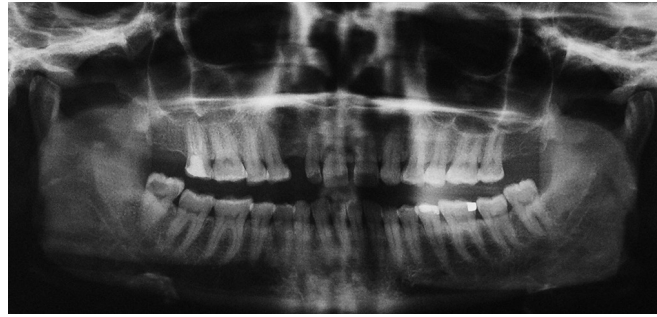
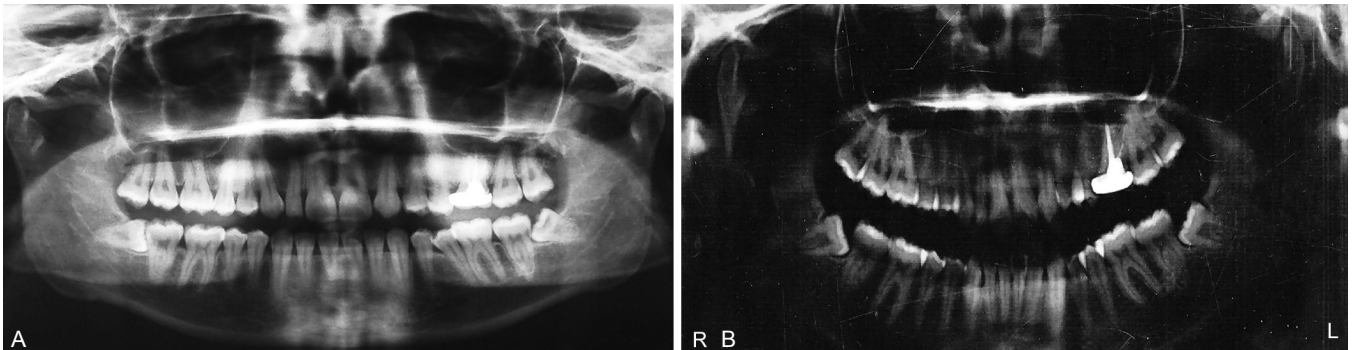


Fig. 4: OPG of patient's mother showing missing 22 and extraction space of 13



Figs 5A to C: Intraoral photographs of patient's sister 1



Figs 6A and B: OPG of patient's sister 1 showing missing 22. (A) Current OPG; (B) Preorthodontic treatment OPG



Figs 7A to C: Intraoral photograph of patient's sister 2 showing retained 55,63,65,75 and extraction space of 85, unerupted 12 and 22



Fig. 8: OPG of patient's sister 2 showing missing 12,22, and 45

was from the maternal side. With regard to the association of the number and type of missing teeth, only two family members exhibited a similar pattern of hypodontia. Both mother and the eldest daughter had missing maxillary lateral incisor of the left side with microdontia of right maxillary lateral incisor. The male member who was the only son in the family had missing maxillary lateral incisor of right side only with microdontia of left maxillary lateral incisor and his second sister had both the maxillary lateral incisors missing along with right

mandibular second premolar. However, this could be explained on the findings of a previous study in which bilaterally symmetrical pattern of tooth agenesis was found, except maxillary lateral incisors where the left one is more frequently missing than the right one.¹⁷ As in this case, at least one missing tooth was observed in both the males and females of the family, it was suggestive of an autosomal dominant pattern of inheritance. However, the dissimilar pattern of hypodontia among the family members could be explained on the basis of incomplete penetrance and variable expressivity.

In recent years, mutations in genes have been identified in human pedigrees with familial hypodontia or oligodontia. Some of the genes identified are *MSX1*, *PAX9* and *AXIN2*.⁶ Current evidence suggests that *MSX1* and *PAX9* genes interact during tooth development.^{18,19}

Hypodontia in young patients can cause esthetic, functional and psychological problems. Depending on the severity of hypodontia, treatment of these patients can vary from absolutely no treatment to extensive treatment. However, treatment usually requires a multidisciplinary approach which may include prosthetic, orthodontic and adhesive restorative techniques or a combination of these techniques.

CONCLUSION

Multiple missing teeth can be diagnosed easily, but what is more crucial is the detection of single or two missing teeth. Since heredity plays an important role in the etiology, the possibility of its occurrence in other family members and in future generations should always be kept in mind. Early recognition is vital for proper treatment planning to prevent malocclusion and other orthodontic complications. Treatment of hypodontia is important to improve esthetics, function and psychological problems and is often facilitated through a multidisciplinary approach.

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