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Abstract

Oligodontia is a congenital absence of one or more teeth, which has familial abnormality, and attributable to various mutations or polymorphisms of gene often associated with malformative syndromes. The present case report, is a rare case of non-syndromic oligodontia in a 8 year old girl with missing 14 permanent teeth buds excluding third molars in mixed dentition, a very rare situation. Mutations in MSX1 and PAX9 have been described in families in which inherited oligodontia characteristically involves permanent incisors, lateral incisors, premolars and molars. Our study analyzed one large family with dominantly inherited oligodontia clinically and genetically. This phenotype is distinct from oligodontia phenotypes associated with mutations in PAX9. Sequencing of the PAX9 revealed a novel mutation in the paired domain of the molecule. PCR and sequence analysis of the PAX9 exon 2 revealed two mutations. Comprehensive interdisciplinary treatment planning with good coordination and timing of the individual treatment phases are presented.

Key Words: MSX1, Tooth development, PCR

Introduction

Agenesis of one or more teeth is one of the most common of human developmental anomalies [1,2]. Oligodontia is a very rare condition, has a population prevalence of 0.03% to 0.07% and occurs most frequently in females at a ratio of 3:2. The most frequently missing teeth are the maxillary lateral incisors, followed by the mandibular second premolars and the mandibular central incisors [3,4].

The etiology of tooth agenesis may vary from physical obstruction or disruption of the dental lamina, space limitation and functional abnormalities of the dental epithelium or failure of initiation of the underlying mesenchyme [4]. It may also occur as part of a systemic genetic syndrome [5], or can also be due to an isolated condition (nonsyndromic oligodontia) like mutation in LTBP3 [6], or mutation in the homeobox gene MSX1 or paired domain transcription factor PAX9 [7].

The absence of permanent teeth may cause several clinical problems and the inconvenience to patients will vary, depending on the age, specific teeth and the number of teeth that are missing. This clinical report describes a rare case of non-syndromic oligodontia in a eight years old girl in mixed dentition stage who had problems with aesthetics, mastication, and phonation. Since, the therapeutic concept of oral rehabilitation by multi-disciplinary approaches are decisive for a successful treatment outcome an early diagnosis, and comprehensive treatment planning with good coordination and timing of the individual treatment phases are presented.

Case Report

A 8 year-old female patient (proband) reported to the department of pediatric dentistry with a chief complaint of multiple decayed teeth in lower left quadrant. The patients past medical history and the family history were not contributory. It was patient’s first visit to a dentist. Extra-oral examination revealed no abnormalities of the skin, hair or nails. Intraoral examination, revealed grossly decayed teeth in relation to 54, 65, 84 and hypoplastic teeth in relation to 64, 74 and 55. A few retained carious deciduous teeth were present in relation to 53, 55, 65, 81, and 85 with huge midline diastema. The proband (III:9) Orthopantomographic (OPG) examination revealed agenesis of eighteen permanent teeth including third molars. The missing teeth were 12,13,15,17,18,22,23,25,27,28,33,35,36,38,43,45,46,48 (Figure 1). The panoramic

Figure 1. OPG of proband.
Radiography also revealed a few developing permanent teeth in relation to 14, 17 and 27. The teeth present were normal in size, shape and color and not associated with any periodontal disease. There was lack of development of mandibular alveolar bone height. Queries revealed missing teeth were not extracted and were absence since childhood. Family history was taken which revealed even her brother and sister having a similar problem. Proband's father (II:7) aged 50 years did not have any missing teeth and mother (II:8) of age 38 years had retained teeth in relation to 63, and congenitally missing teeth in relation to 12,22,18,28,38,48 (Figure 2). Proband brother (III:7) of age 17 years was examined clinically and radiographically which revealed retained primary teeth in relation to 52,53,62,71,72,73,75,81,82, and congenitally missing permanent teeth 12,13,22,31,32,33,35,41,42,45,18,28,38 (Figure 3). Proband sister (III:8) of age 13 years was examined clinically and radiographically, which revealed root stumps in relation to 74 and congenitally missing teeth in relation to 12,18,22,28,38,48 (Figure 4).

Once it was confirmed that the patient and elder brother were affected with oligodontia, elder sister and mother with hypodontia, genetic evaluation was planned for the entire family.

Familial pedigree chart was drawn. Proband's both parents side pedigree chart was drawn separately (Figure 5).

**Pedigree Chart**

Proband's three generation familial pedigree chart was drawn. Proband's both parents side pedigree chart was drawn separately. Proband's grandparents of both side were no longer lived, so history of missing teeth could not be assessed, excluding them the entire members in the family (25 members) were examined clinically. Except for the proband, her siblings and her mother, no one else in the entire family had any history of congenitally missing teeth.

Clinical phenotypes of affected proband, mother and siblings are tabulated.

### Table 1

Once it was clinical confirmed of congenitally missing permanent, we were curious to find out genes which was responsible for agenesis of teeth. So in this case all the family members of proband were subjected to genetic evaluation.

### Genetic Evaluation

The entire procedure was explained to the family members and an informed consent was obtained. 5ml of venous blood was with-drawn from all the family members of proband to extract DNA and was transported to Vittal Mallaya Research Foundation for further genetic evaluation.

PCR and sequence analysis of the PAX9 exon 2 revealed two mutations (T to C) at 48th and 64th nucleotide. These two mutations alter the translated protein sequence.
were extracted under local anesthesia. Band and loop space maintainer was given in relation to primary maxillary right first molar (54) and primary mandibular right first molar.

ionomer cement in relation to 55 and 85. Hypoplastic teeth in relation to 64 and 74 were restored with stainless steel crowns. Grossly decayed teeth in relation to 54, 65 and 84

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Table 1. Clinical phenotypes of affected proband, mother and siblings. ♦ represents congenitally missing teeth.

**Figure 6.** Sequences of PAX9 exon in proband’s sample. The mutations are highlighted.
(84). Mayen’s space maintainer was given in relation to primary maxillary left second molar (65), and the patient is under observation and regular follow up till the eruption of succedaneous teeth. Fluoride application is done on a regular basis.

After pediatric management, Orthodontic therapy should be followed, which presently involves, midline diastema closure of the missing permanent maxillary lateral incisors. Alignment of lower permanent anteriors using Sectional Oven Niti wire (0.016), followed by 19x25 Niti wire and then 19x25 stainless steel wire. Space maintainer to be continued till permanent tooth eruption. Primary canines and molars are maintained in position till their exfoliation. Evaluation of root of primary molars is done for any resorption every 6 months. After completion of growth i.e. after 18 years of age replacement of 12 and 22 with fixed prosthesis. Evaluation of bone distal to permanent first molars done for replacement of second molar using implants.

Following orthodontic treatment prosthetic management includes giving removable prosthesis with periodic checkup of the prosthesis every year, needs to be done to evaluate the fit of the prosthesis in the growing child. Once the patient reaches the age of 18 years and above a permanent fixed prosthesis can be placed based on the amount of edentulousness exists which may include implant retained prosthesis.

**Orthodontic consideration**

The orthodontic treatment of patients with congenitally missing laterals and canines is inconclusive as to whether to close the spaces left by the missing lateral incisors orthodontically or to open or maintain spaces for prosthetic (FPD) replacement or implants [12]. One report advocates, opening or maintaining the space for prosthetic replacement or implants in order to achieve a better occlusion and less flattening of the facial profile is obtained [13].

As in our case after pediatric management, orthodontic therapy is planned accordingly, which involves closure of the midline diastema in relation to 11,21, and alignment of lower permanent anteriors using Sectional Oven Niti wire (0.016), followed by 19x25 Niti wire and then 19x25 stainless steel wire. Space maintainers which is already placed to be continued till permanent tooth eruption. After completion of growth i.e after 18 years of age the missing teeth will be replaced with fixed prosthesis or using implants.

**Prosthetic consideration**

It is a general rule that the final prosthetic solution (fixed/implants) should be avoided until the end of growth and development. Symptomatic treatment in the form of provisional prosthetic replacement like removable partial dentures can be considered till the age of 18 years. Removable prosthesis should be checked periodically, to be done to evaluate the fit of the prosthesis in the growing child.

After complete eruption of the teeth, repositioning and minor correction to best of esthetics and function should be considered. Vertical dimension and occlusal plane should be evaluated before the final prosthetic replacement. Any centric and eccentric interferences should be removed. Once the patient reaches the age of 18 years and above a permanent fixed prosthesis can be placed based on the amount of existing edentulous space which may include implant retained prosthesis.

In our case considering the age of the child and the status of the dentition i.e primary and permanent teeth, necessary treatments like oral prophylaxis, restorations, stainless steel crowns and space-maintainers was preferred. As the child grows a multidisciplinary approach is essential to achieve better aesthetics and function in such cases.

**Conclusion**

In conclusion early diagnosis, and comprehensive treatment planning with good coordination and timing of the individual treatment phases are decisive for a successful treatment outcome. The parents should be educated about probable future treatment options for their growing child to prevent future functional and aesthetic problems. The inconvenience to patients will vary depending on the specific teeth and the number of teeth that are missing. It is necessary for us to monitor this case to achieve better results in the future.

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