

Nonsyndromic Oligodontia in Permanent Dentition: Three Rare Cases

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Abstract: *Oligodontia is the congenital absence of six or more than six teeth in either permanent or primary dentition. Because of the missing teeth in these patients esthetic, functional and psychological problems may arise. This article reports a three rare cases of non-syndromic oligodontia.*

Key words: *oligodontia, hypodontia, severe partial anodontia*

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I. Introduction

Dental agenesis is the most common developmental anomaly in humans, often presenting a significant clinical problem. It is classified according to the number of missing permanent teeth excluding the third molars. Hypodontia is used to describe the absence of one or few teeth, Oligodontia is used for agenesis of numerous teeth (more than six teeth) excluding the third molars and anodontia is the extreme of oligodontia where there is total absence of any dental structure.[1] Oligodontia is also known as partial anodontia, severe or advance anodontia.

Some of them also refer this as selective tooth agenesis. According to different authors, the frequency of hypodontia is 1- 10% and oligodontia 0.1-0.9%.

Anodontia occurs very rarely (17 cases were described over the last 50 years).

Oligodontia may occur as a part of a genetic syndrome, as a non syndromic isolated familial trait, as an infrequent finding or as an isolated condition that has been linked to mutations of the MSX1 and PAX9.[3,4,5]

The third molar (M3) represents the tooth most affected with agenesis, 2, 3, having a prevalence rate of 20.7%. In contrast, permanent second molar (M2) agenesis is a rare occurrence, found in only 2 of 6,000 consecutive orthodontic patients (0.03%)[6]. Excluding the third molars, the prevalence rate of tooth agenesis is reported as 4.3 to 7.8%[4, 6]. The mandibular second premolar (MnP2) is the tooth most often absent, with a relative frequency of 2.2 to 4.1%[4, 5]. In fact, the MnP2 is highly variable developmentally. Besides the high prevalence of agenesis, the MnP2 often shows significantly retarded development, especially when there is agenesis of other permanent teeth[6]. Despite the fact that the mean initial calcification age for MnP2 is 3 years (varying from 2y3m to 3y7m)[7], its development can be suppressed until 6 years[8], and some published reports show radiographic appearance of the MnP2 after the age of 9 and even at 13 years old[9, 10]. In addition, the MnP2 accounts for approximately 24% of all impacted teeth, excluding the third molars[11]. The most frequent malposition reported for the unerupted MnP2 is distoangular development, with a prevalence rate of 0.2% in dental clinic patients[12]. This malposition was found to be associated with agenesis of the contralateral MnP2.[11,12]

Molecular studies have revealed that the instructive and permissive tissue interactions during mouse tooth development described above are mainly mediated by growth factor signalling. Development from initiation to eruption is governed by a sequential and reciprocal signalling process rather than simple one-way messages. The signalling involves all major signalling pathways, including transforming growth factor b (TGFb), fibroblast growth factor (FGF), sonic hedgehog (Shh), anhidrotic ectodermal dysplasia (Eda), and epidermal growth factor (EGF) signalling, and studies with mouse mutants have shown that they are needed simultaneously during critical stages of development.[9]

Msx1 and *Pax9* are transcription factors intimately involved in the genetic networks regulating tooth development. *Msx1* contains a homeobox which binds to specific target sequences in the DNA but is also capable to proteins interaction. *Msx1* has often been considered rather as a repressor than activator of gene expression. *Pax9* belongs to the paired-box containing transcription factor family, and is one of the earliest mesenchymal markers of the future tooth forming positions in mouse. *Pax9* is regulated by epithelial signals, especially FGF8, and it apparently regulates reciprocal signalling from the mesenchyme. In mice with hypomorphic *Pax9* mutations, a partial failure of tooth development was observed, affecting in a dose-dependent manner the third molars and incisors and to a smaller extent the other molars. The ameloblast differentiation and dentinogenesis were also affected. [8,9,10]

It has been suggested that the key role of *Msx1* and *Pax9* is to facilitate the bud to cap stage transition. There is signals emanating from the epithelium and mesenchymal during tooth development and molecular regulation . Mesenchymal *Msx1* expression is initially activated by the epithelial bone morphogenetic protein 4 (BMP4) signal, and needed for a reciprocal BMP4 signal from the mesenchyme. BMP4 and *Msx1* thus form an autoregulatory loop. BMP4 signal to the epithelium is crucial for the formation of the epithelial signalling centre, the enamel knot, and the arrest of the development in *Msx1* null mutant teeth can be rescued by external BMP4 or transgenically activated BMP4 expression. The expression of *Pax9* is apparently needed to maintain and, by the synergism with *Msx1*, to enhance this loop and also needed later in tooth development. [8,9,11,12]

This article aims. In this report, is to demonstrate the isolated hypodontia that paternally exists in three siblings. The diagnosis of hypodontia should be performed as early as possible in order to prevent aesthetic and functional problems in dentition.

II. Case Reports:

Case report; 1

A 13-year-old female patient reported to my private Pediatric Dental Clinics reporting absence of some teeth. Through a digital panoramic radiograph the existence of multiple agenesis of permanent dentition was revealed. In the radiograph agenesis of tooth 15, 25,34, 35, 44,45 (Fig. 1) was identified, with a small dimension of maxilla. Also, a slight condylar asymmetry with a small size and a slight stylohyoid ligament ossification was noted in the left side. After this, a foot radiograph was taken to determine if the condition had a relation with an osteopetrosis; however, normal findings were noted. During anamnesis the patient reported she had no trauma history, previous tooth extraction, orthodontic treatment or complications during pregnancy or birth. The patient's mother informed that there was no history of syndromic or systemic disease. At general examination no alterations or systemic diseases were identified, with facial symmetry, no palpable lymph nodes and both jaws were normal.

Clinically, in the intraoral examination no caries and the absence of the same teeth were observed with tooth rotation of 13, 23 and 43. No presence of periodontal disease was noted.



Fig: 1. Orthopantomogram of the patient Case 1

The patient was examined to rule out syndromes associated with oligodontia. She was normal in his facial appearance and did not show any physical or skeletal abnormality. Radiological examinations of the clavicles, vertebral skeleton, skull and chest were found to be normal.

Ophthalmological and neurological examination of the patient revealed no pathological symptoms and showed no signs of mental retardation. Hematological and biochemical findings were within the normal limits.

Case report; 2

An 8-year-old girl reported to the to my private Pediatric Dental Clinics the chief complaint of missing lower front teeth. The parents had observed this condition since early childhood but did not seek any dental consultation, and there was no history of any previous trauma or extraction. Past medical and family histories were



Fig.2 - Orthopantomograph of the patient case2

Noncontributory. On extraoral examination a well balanced face, with an apparent eye defect and a convex facial profile was observed. Intraoral examination revealed (i) mixed dentition stage, with (ii) mesial step molar relationship, (iii) root stumps of 52, 61, 62, 73, 84, (iv) missing 41, 45, 31, 33 and (v) multiple carious teeth . The alveolar ridge in the mandibular anterior region was typically knife edged , suggesting absence of mandibular anterior teeth. To ascertain the provisional diagnosis, an orthopantomograph was advised and revealed no evidence of development of 31, 32, 33, 41, 42, 45 (Figure 2). Parents were informed about the agenesis of six mandibular permanent teeth.

Case report; 3

A 13 year old boy reported to the my private Pediatric Dental Clinics for a routine dental check-up. His past medical history was non-contributory and family history revealed that he was born to non-consanguineous marriage with normal delivery and no one in his family have congenitally missing teeth. The patient had no history of trauma or extractions. Extra oral examination revealed a face with normal facial profile and normal skeletal dental base relations . During clinical examination, maxillary central incisors were conical in shape along with bilateral peg shaped maxillary lateral incisors. The deciduous right mandibular central incisor and maxillary canines, first and second molars were retained and permanent mandibular incisors weremissing. Radiographically, in Orthopentomogram (OPG) all four permanent second molars and permanent mandibular central and lateral incisors were missing (fig 3).



Fig.3 - Orthopantomograph of the patient case3

III. Discussion

Congenitally missing if it has not erupted in the oral cavity and is not visible in a radiography. In general hypodontia is the term most frequently used when describing the phenomenon of congenitally missing

teeth. Hypodontia is an anomaly that may result in dental malpositioning, periodontal damage, lack of development of maxillary and mandibular bone height and has significant psychological, aesthetic and functional consequences. Hypodontia and oligodontia are classified as isolated or non-syndromic, where as hypodontia/oligodontia and syndromic.[1,6]

The prevalence of permanent tooth agenesis ranges between 1.6% and 9.6%, and the prevalence of deciduous tooth agenesis is lower, ranging between 0.5 % and 0.9 % [6]. The biologic basis for the congenital absence of permanent teeth is partially explained by the failure of the lingual or distal proliferation of the tooth bud cells from the dental lamina. The causes of hypodontia are attributed to environmental factors such as irradiation, tumours, trauma, hormonal influences, rubella, and thalidomide or to hereditary genetic dominant factors, or to both. Familial tooth agenesis is transmitted as an autosomal dominant, recessive, or X-linked condition. Affected members within a family often exhibit significant variability with regard to the location, symmetry and number of teeth involved. Residual teeth can vary in their size, shape or rate of development and the permanent dentition is more affected than the primary dentition.[7]

Oligodontia can be found as an isolated nonsyndromic trait or as a part of a syndrome, such as, ectodermal dysplasia, Down syndrome, incontinentiapigmentii and rieger syndrome. Mutation in the transcription factor MSX1 and PAX9 have been identified in families. The factors were demonstrated to be associated with isolated non-syndromic oligodontia. Recent studies have shown that mutation in EDA gene could result in non-syndromic oligodontia [13]. These isolated forms may be sporadic or familial. Familial tooth agenesis can be result of single dominant gene defect, a recessive or X-linked [8,9] .

Grahnen has suggested that tooth agenesis is typically transmitted as an autosomal dominant trait with incomplete penetrance and variable expressivity[14]. In his sample the penetrance was higher when the proband of the family had more than 6 missing teeth .[9]

Congenital agenesis of teeth can create dental and facial disfigurement, which can lead to social withdrawal, especially in adolescent years. Treatment in all the presented three cases includes multidisciplinary team approach of pedodontists, orthodontists, oral and maxillofacial surgeons, and prosthodontists to restore aesthetics, functional, and psychological reasons depending on the severity of the condition and patient's perceived need for care. Factors to be taken into account before treatment planning includes age of the patient, number and condition of retained teeth, number of missing teeth, condition of supporting tissues, the occlusion, and the interocclusal space.[1,6]

Oligodontia condition should not be neglected as it may result in various disturbances like abnormal occlusion, altered facial appearance which may cause psychological distress, difficulty in mastication and speech. Treatment depends on extent of hypodontia and should consist of interdisciplinary approach. Therefore early diagnosis is important in such conditions. Case of tooth agenesis should be recorded with complete clinical history including medical and radiological investigations to rule out any syndrome .[15]

In the cases studies the remaining teeth were carious, had malposition, polidistema and malformation. No definite etiologic relationship has been found between hypodontia and systemic diseases or endocrine disturbances

Oligodontia is frequent finding in many syndromes, but in this caseS it was not associated with any syndrome which is a rare finding. So as an oral physician if we come across any case having multiple congenital missing teeth it may not be always associated with multiple other abnormalities as seen in syndromes.

Patient suffering from oligodontia may have severe psychological, esthetic, functional problems and early diagnosis and treatment is very important. Treatment approach has to case specific and depends on condition of primary predecessor, number of missing teeth, status of occlusion and patient and parent's preferences. Options include orthodontic therapy, implants, removal partial prosthesis, fixed prosthesis, over dentures and indicated depending on the type of condition. Treatment not only improves speech and chewing function but also has psychological implications that may greatly help in regaining the self-confidence[16].

IV. Conclusion

The concept of early diagnosis of these patients becomes more important. When a case of tooth agenesis is seen, the presence of the anomaly should be recorded with a complete clinical history including panoramic radiograms and models, for proper phenotype characterization prior to any surgical or orthodontic treatment. There are a number of options available to restore space generated by missing teeth. Dental treatment can vary depending on the severity of the disease and generally requires a multidisciplinary approach.

Treatment options include orthodontic therapy, implants, adhesive techniques, removable partial prostheses, fixed prostheses and over dentures and they are indicated depending on the type of condition. Most cases require the construction of a partial denture as an intermediate procedure before fixed prostheses are constructed. Treatment not only improves speech and chewing function but also has psychological implications that may greatly help in regaining self-confidence.

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