Case Report

Non-Syndromic Oligodontia: A Case Report
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Abstract
Oligodontia is a rare and severe degree of dental agenesis, characterized by the congenital absence of more than six dental elements excluding the third molars. The prevalence rates of this condition are around 0.3% and the most frequently missing teeth are the maxillary lateral incisors and the mandibular second pre-molars. This condition may be associated to a syndrome or occur as a non-syndromic form, in familial traits or sporadic cases. It has been shown that the congenital absence of dental elements are given to defects in relate genes, mainly the PAX9 and MSX1 genes, which are indispensable for the teeth development. Several authors stand up for the hypothesis of the human dental reduction of size and number is given to evolutionary factors, specifically, changes in dietary patterns. This paper presents a case of oligodontia in a 20 years old male patient diagnosed with oligodontia due to the congenital of eleven dental elements.

Keywords: Dentition; Agenesis; Diagnosis; Hypodontia; Odontogenesis; Dental Practice Patterns

Introduction
Tooth agenesis or congenitally missing teeth has been reported as the most prevalent anomaly of the craniofacial development in humans, representing a challenging problem to the clinical management, both functional as aesthetic. This teeth absence may be classified depending on the number of dental elements missing as: hypodontia, when there are one to six missing teeth, excluding third molars, oligodontia, when there are more than six dental elements missing, also excluding third molars and anodontia, when occurs the general absent of teeth development.

Oligodontia is a rare and severe degree of dental agenesis, showing prevalence rates around 0.3%, presenting slight predilection for the female gender, and previous studies have shown that the prevalence of the type and also the number of teeth absence may vary according to the population studied. This condition is mostly common in the permanent dentition, and the diagnosis is usually based on radiographic evidence and clinically on the delayed eruption of the permanent teeth. Nevertheless, several other alterations may be noticed during the diagnosis process, including reduced size and form of teeth, delayed growth of the alveolar processes, deficiency of teeth eruption, persistent deciduous teeth, taurodontism, false diastema, and deep overbite. Speech and masticatory disorders may also be noticed. The rare cases of oligodontia may occur associated to a recognized genetic syndrome condition or, more commonly, as an isolated trait, in these cases, the tooth agenesis can be result of a familial trait or sporadic in nature. However, in both syndromic and non-syndromic cases, tooth agenesis is associated to expression disturbs of relate genes, particularly the PAX9 and MSX1 genes, which plays key roles in early stages of tooth development.

Due to the complex necessity to rehabilitate functional and aesthetically affected patients, clinical management of oligodontia requires a multidisciplinary approach, which might involve orthodontic, prosthetic treatment including implants, and speech therapy. This paper reports a rare case of isolated sporadic oligodontia in a 20 years old patient, missing eleven permanent teeth.

Case Report
A 20 years old male patient, Japanese descendent, reported for routine examination. Medical history was non-contributory. Dental history revealed that the patient was diagnosed with dental agenesis as a child, when he appeared to private orthodontic treatment. The primary teeth which had no successor were maintained, and after the orthodontic correction, the anterior teeth with diastema were corrected with composite resin. The patient brought his dental documentation, containing images of the clinical aspect of his dentition at age of six years old (Fig 1) showing the severe overbite of the element 61, and the absence of the element 62. This documentation also contained two panoramic radiographies. The
evaluation of the oldest radiography, acquired when the patient was six years old (Fig 2A), reveals several missing dental germs of the permanent dentition: maxillary lateral incisors, maxillary and mandibular second pre-molars, maxillary right second molar and maxillary and mandibular third molars. The case was followed up and the evaluation of the panoramic radiography corresponding to the age of eight years old (Fig 2B) confirms the congenital absence of the related dental elements, as the present germs were normally evolving and erupting, and also reveals the presence of a supernumerary tooth in the region of 48, which was surgically removed as related by the patient.

Figure 1: Clinical aspect at 6 years old.

Figure 2: Radiographic aspect at 6 years (A) and 8 years (B) old.

The clinical examination (Fig 3) revealed the presence of the primary dental elements 53, 55, 65, 75 and 85, the missing bilateral permanent maxillary lateral incisors were placed by the permanent maxillary canines, leaving diastemas, which were corrected with composite resin. A new panoramic radiography was acquired (Fig 4) revealing no impacted teeth and corresponding to the clinical aspect before mentioned, the roots of the primary teeth are still implanted on the jaws, with no signs of ankylosis or reabsorption. All permanent and primary teeth are correctly positioned on the dental arch, except for the deviation of the midline. To sum up, the present teeth are: 11, 13, 53, 14, 55, 16, 21, 23, 24, 65, 26, 27, 31, 32, 33, 34, 75, 36, 37, 41, 42, 43, 44, 85, 46 and 47; and the missing teeth are: 12, 22, 15, 25, 35, 45, 17, 18, 28, 38 and 48. No occlusion or speech disturb were related, due to early orthodontic correction, the primary teeth presents were healthy and did not presented with mobility. Aesthetically the patient did not show interest in treatment for now. Based on the absence of seven permanent teeth, excluding the third molars, the patient was diagnosed with oligodontia. Including the third molars, the total of missing dental elements is eleven.

Figure 3: The clinical aspect.

Figure 4: Actual radiographic aspect.

Discussion

Dental agenesis is a term widely used to describe the failure of tooth to develop, causing definitive absence of one or more teeth, which is not such a rare trait, with prevalence rates varying from 2.8% to 14.9% in population studies excluding the third molars.\(^8\)-\(^10\) According to some authors, when including the absence of third molars, the prevalence of dental agenesis may increase to values around 49%.\(^11\) and according to review by Pani (2011)\(^2\), up to 25% of the population may lose at least one third molar. Oligodontia describes the congenital absence of more than six dental elements\(^2\), being a rare trait, showing prevalence rates around 0.21 to 1.4%\(^12,13\), with a slight predilection to female gender, what does not lay with this case, which in oligodontia occurred in a male patient, and may vary according to the different ethnic groups studied, being the Japanese the most commonly affected\(^1\), what does
corroborate with this case, once the patient is Japanese descendant.

The pattern of absence in this case was compatible with epidemiological studies that showed that the most commonly congenital missing teeth are the third molars, followed by the upper lateral incisors and the mandibular second pre-molars, and is also compatible with other cases of oligodontia. The case presented here also shows an extremely rare situation termed Concomitant Hypo-hyperodontia (CHH), when occurs the simultaneous dental agenesis and supernumerary dental elements in the same individual, the prevalence rate of CHH is around 0.33% among orthodontic patients. In this case, the supernumerary tooth, located in the region of mandibular right second molar was surgically removed when the patient was 10 years old as related by himself. Oligodontia might occur associated to a syndrome or, more commonly, occur as a non-syndromic disturb of craniofacial development, as a familial trait or sporadic cases, as the case presented on this paper, which on the familial historic of the patient did not revealed any case of any degree of dental agenesis. However, the occurrence of dental agenesis is always associated to expression disturbs in related genes, and might be an autosomal dominant, autosomal recessive or X-linked trait. Mutation spectra of non-syndromic congenital missing teeth in humans is due to defects in two such genes that encode transcription factors, the MSX1 and PAX9 genes.

The MSX1 gene is located in the chromosome 4 and is involved in several epithelium-mesenchyme interactions during embryogenesis. It's highly expressed in the mesenchyme of the dental buds, particularly during its initial stages. The PAX9 gene is present in the chromosome 14, and belongs to a family of genes that encompasses a group of transcription factors which act during the initiation of the embryo's development. The expression of the PAX9 is necessary for dental mesenchyme to condense around the tooth bud epithelium. The respective protein products of the PAX9 and MSX1 play an important role in the maintenance of mesenchymal bone morphogenetic protein 4 (Bmp4) expressions, which drives the morphogenesis of the dental organ. Up to now, 16 mutations in the PAX9 and 11 mutations in the MSX1 have been identified in humans, thus, once there is an expression defect of these genes, there might be arrested of tooth bud development, leading to agenesis. Studies has also shown that although, apparently, both of these genes interact to each other on the tooth development, mutations on PAX9 and on MSX1 genes may present distinct clinical features regarding the pattern of the dental absences, so that the MSX1 is more associated to agenesis of the anterior teeth while the PAX9 is more commonly associated to the congenital absence of the posterior teeth. The case presented here, showing the congenital absence of anterior and posterior teeth is possibly given to mutations in both of these genes, while both of them have been shown mutated in patients with diagnosis of oligodontia.

The congenital absence of dental elements is categorically defended by some authors as an evolutionary adaptation, Calgano and Gibson (1988) presented the "probable mutation effect" which defends that structures no longer functional owning to ecological or cultural changes will experience a relaxation of selection pressure, allowing an accumulation of mutations in the population that inevitably will result in the reduction in the size or the loss of the concerned structure, the authors also commented that the best illustration of structural reduction in man is in the face, due to the increasing deployment of tools and techniques of food preparation, large teeth are no longer needed. In review by Emes et al., (2011) the human masticatory system, just like other anatomical features of our specie, has also evolved during the history of man, and also commented that due to dietary changes undergone over the years, our specie has passed by changes in the size of the jaws and teeth, and also in the number of teeth. Therefore, the dental agenesis may be considered as an evolutionary trend and any of genes involved on odontogenesis may have participated on the dental evolution. Besides, according to review by Liu (2011), most frequently missing teeth is the most posterior teeth of each group – among them incisors, pre-molars and molars, what resembles with the case here presented, hypothesizing that most frequently missing teeth were "vestigial organs" that does not imply selective advantage to the specie.

The case presented is rare by occurring in a non-syndromic male patient, when it’s a characteristic frequently found in syndromes,
and the prevalence studies has shown predilection by the female gender. It is also important to accentuate the role of early diagnosis, allowing early multidisciplinary approach, which is important to avoid future masticatory or speech disturb. The congenital teeth absent may be considered as an evolutionary trait.

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References

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