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Case Series

Parental consanguinity a possible risk factor for tooth agenesis: A case series

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ABSTRACT

Introduction: Dental agenesis is a commonly encountered dental anomaly that affects more than 20% of the human population. It can be categorized as hypodontia, oligodontia, or anodontia. Oligodontia can occur either as an isolated finding or as a part of the syndrome, and the causative factor can be either environmental or genetic.

Case Presentation: This report describes three cases of isolated, non-syndromic tooth agenesis with single/multiple missing permanent teeth. No other associated clinical features or any systemic involvement were present in these cases, suggesting parental consanguinity as a possible etiological factor.

Conclusion: The purpose of this case report is to promulgate premarital counselling and health awareness programs focusing on the potential consequences of consanguineous marriages and their prevention.

1. Introduction

Developmental anomalies like dental agenesis are commonly encountered in routine dental examinations and affect more than 20% of the human population, consequently causing esthetic, psychological, and functional ramifications. The prevalence rate varies depending on the demographic and geographic profiles.[1] It is seen that developmental anomalies associated with the number of teeth are more common in females and particular races. For example, hypodontia associated with mandibular central incisors occurs commonly in Japanese and Swedish populations.[2]

Dental agenesis can manifest as anodontia, hypodontia, or oligodontia. Hypodontia is a condition whenless than six teeth are congenitally missing, whereas oligodontia involves the absence of six or more teeth excluding the third molars. Anodontia is a condition where all the teeth fail to develop, and it occurs in rare situations.[3,4]

The aetiology of dental agenesis is complex and multifactorial as over 300 genes contribute to it.[5] Congenitally missing teeth can vary genetically and phenotypically. It can occur either in association with hereditary ectodermal- dysplasia, Gardner syndrome, cleidocranial dysplasia, and down's syndrome or in patients with hormonal imbalance. Local factors like metabolic derangements, trauma, drug usage for treatment of malignancies during formative stages, and infections are also contributive.[6–8] The absence of teeth may be unilateral or bilateral, and the most frequently congenitally missing permanent teeth excluding third molars are the mandibular second premolar (3.4%) and the maxillary lateral incisor (2.2%).[9,10] Other dental anomalies that can coexist with tooth agenesis are enamel hypoplasia, primary molar infraocclusion, and palatally inclined or impacted maxillary canines along, with the prominent generalized spacing and rotations of teeth adjacent to missing mandibular second premolars.[11]

The currently available literature showing the association of parental consanguinity and dental anomalies is under-

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explored and almost silent about the potential correlation. Besides, there is also insufficient data on agenesis of bilateral permanent mandibular central incisors.[12,13] This article presents a series of 3 case reports with missing permanent teeth in children with the consanguineous parent.

2. Case Report 1

A 13-year-old girl reported to the Department of Paediatric and Preventive Dentistry, Jamia Millia Islamia, New Delhi, India with the chief complaint of broken front teeth in the lower arch. Intraoral examination revealed the presence of retained deciduous mandibular central incisor in the midline having grade I mobility**.** On meticulous clinical examination and radiographic evaluation, it was noticed that the central incisor was placed on the left side of the midline hence the tooth was diagnosed as 71.

On intraoral examination, we observed that both the permanent mandibular central incisors had not erupted. It was ascertained on further clinical exploration that the second premolars had not erupted while the deciduous second molars were present in both the arches with generalized spacing in between the mandibular anterior teeth. There was no history of any tooth extraction in the past. On extraoral examination patient's skin, nails, hair and were found to be normal in texture and appearance. Also, no sweating abnormality was detected which ruled out the associated syndromes with oligodontia. Her medical record revealed a normal birth with no history of trauma or infection to the anterior teeth region, absence of any systemic compromise, or any other relevant datum. The patient was the only child born to consanguineous parents and no other dental anomaly found in the family.

On panoramic radiographic examination, the absence of all four second premolars and permanent mandibular central incisors were confirmed. Along with this, evidence of initial calcification was seen in the third molar region of the maxilla while there was no evidence of the third molar crown or follicle in the mandible. Considering the age of the patient, these findings were unusual. (Figure 1). The thyroid profile of the patient revealed normal thyroid profile parameters. Based on the subject's history, clinical observations, and radiographic findings, this case was diagnosed as non-syndromic Oligodontia.

3. Case Report 2

An eight-year-old male child reported to the department with a chief complaint of several decayed teeth in the upper and lower back teeth region of the mouth. The child was in good health and medical history did not reveal any systemic illness. The child was born to consanguineous parents. The mother reported uneventful pregnancy and no significant family history. The extraoral assessment showed neither facial asymmetry nor skeletal malocclusion. The child was

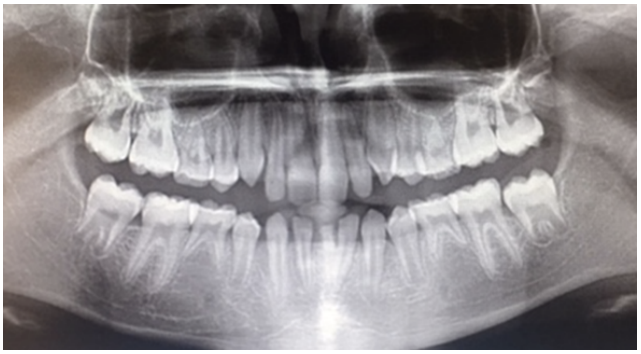

Fig. 1: Orthopantomogram of Case 1 showing congenital absence of all four second premolars (15,25,35,45) & permanent mandibular central incisors (31& 41)

examined with specific regard for hair, ears, nail, eyes, all of which gave off an impression of being normal. No sweating abnormality was reported by the patient which ruled out the possible oligodontia associated syndromes. Intraoral examination revealed decayed maxillary and mandibular primary molar teeth (54,55,64,65,74,75,84,85). On panoramic radiographic examination, the absence of underlying tooth bud of second premolar (15) was accidentally found**.** (Figure 2) The patient was diagnosed as a case of non-syndromic hypodontia.

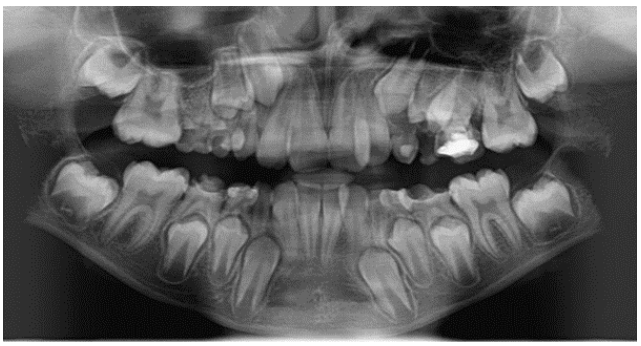

Fig. 2: Orthopantomogram of Case 2 showing congenital absence of second premolar (15)

4. Case Report 3

A seven-year-old male child reported to the department with a chief complaint of decayed teeth in the upper and lower back teeth region of the mouth. His medical history was non-contributory. The child was born to consanguineous parents. On extraoral examination patient's skin, nails, hair and were found to be normal in texture and appearance. Also, no sweating abnormality was detected which ruled out the associated syndromes with oligodontia. Intraoral examination revealed retained primary central incisor (51), palatally erupting permanent central incisor (11) and multiple root stumps of maxillary and mandibular primary

molars (54,55,64,65,74,74,84,85).

Panoramic radiographic examination revealed a bilateral absence of underlying tooth bud of mandibular second premolars (35,45). (Figure 3) Based on the patient's history, clinical and radiographic examination, this case was diagnosed as non-syndromic hypodontia. Parents were counselled for the bilaterally missing permanent tooth buds and various age-appropriate treatment options. A multidisciplinary treatment plan was carried out for the rehabilitation of the patient. The patient is kept under careful observation and time appropriate guidance will be provided in follow up visit.

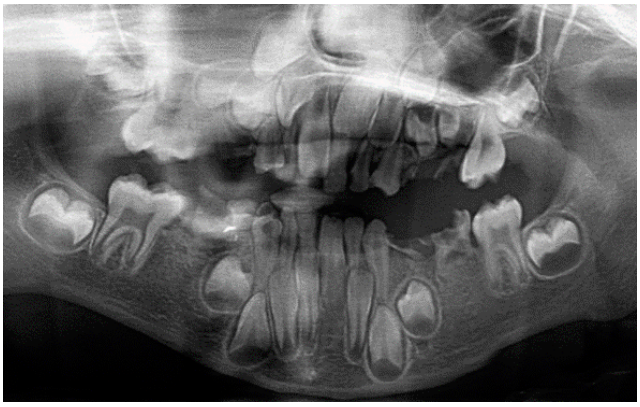

Fig. 3: Orthopantomogram of Case 3 showing congenitally missing bilateral mandibular second premolars (35,45)

5. Discussion

Oligodontia, a form of tooth agenesis, is an unwonted dental anomaly marked by the congenital absence of 6 or more teeth in primary, permanent, or both dentitions. This condition can occur as a part of a genetic syndrome, or as a non-syndromic familial trait or as a sporadic entity.[14] Oligodontia has been reported in 63% of subjects with Down syndrome in the dental literature.[15] However, the non-syndromic form of oligodontia or hypodontia is more frequent, and the biological rationale is explained evidently as the failure of the proliferation of tooth bud cells from dental lamina.[16] The permanent dentition is more frequently affected than the primary dentition. In the primary dentition, the prevalence of tooth agenesis is reported to be 0.5%-0.9%.[14]

The causative factors responsible for dental agenesis can be either environmental or genetic. The environmental factors include trauma to the alveolar processes, infectious diseases like rubella, Candida, etc., endocrine disorders like hypoparathyroidism, psuedoparathyroidism, radiotherapy, chemotherapy, intrauterine disorders, drugs such as thalidomide and disturbances in the innervations of the jaw.[8,17,18] In all three patients, based on the anamnesis, the possibilities of environmental factors were ruled out.

Non-syndromic oligodontia and hypodontia is inherited as an autosomal dominant trait. Various genes involved are TGF-beta, transcription factors like MSX1 and PAX9, and few genes of the wnt signalling pathway. However, MSX1 and AXIN2 genes are involved in the early stages of odontogenesis and, a mutation in these genes is associated with tooth agenesis. It has led us to the understanding of a wide variety of patterns of agenesis.[5,19,20] Oligodontia or hypodontia as a part of any syndrome may have associated abnormalities in other parts of the body besides dental anomalies. The associated structures may include hair, skin, nails, sweat glands. However, in present cases, no such findings were present, and the patients did not have any other physical characteristics of a syndrome, suggesting them to be cases of non-syndromic oligodontia and hypodontia.

The accurate diagnosis of tooth agenesis is extensively dependent on the anamnesis, clinical and radiographic examination performed by a dentist. In the present case 1, a panoramic radiograph revealed congenitally missing bilateral mandibular permanent central incisors along with all the four second premolars. Along with this, it was astounding to observe that there was no evidence of third molar crowns or follicles in the mandible whereas, signs of initial calcification were present in the maxilla. These findings were unusual as literature states that the crown completion of third molars would be completed by the age of 13 and 14 years for boys and girls respectively.[21] The commonest congenitally missing teeth reported are mandibular second premolars followed by maxillary second molars and maxillary lateral incisors.

Interestingly, in this case, mandibular central incisors were missing bilaterally which brings the need for reporting this case since agenesis of bilateral permanent central incisors is a rare condition with a reported prevalence of only 2.2%.[22]

Several factors must be taken into consideration for treatment planning of such cases. Among them, the age of the patient is of major importance, which plays a significant role in determining the treatment outcome. Other conditions that must be evaluated include the number and location of retained teeth, number of missing teeth, presence of caries, condition of supporting tissues, occlusion, and the inter-occlusal rest space.[23] Early detection and treatment of patients with oligodontia generally require an integrated multidisciplinary approach involving pedodontists, orthodontists, maxillofacial surgeons, and prosthodontists. Various treatment options include pre-restorative orthodontics, restoration with removable or fixed partial dentures, rehabilitation with dental implants.

Dentists face many challenges while treating patients with oligodontia and hypodontia. These include space management, uprighting and alignment of present teeth, deep overbite management, and adequate management of

retained deciduous teeth. Space closure is achieved with eruption guidance, while deep bite management requires correction orthodontic bite opening. It is followed by prosthetic rehabilitation using removable dentures or the use of autotransplantation techniques. Any remaining spaces can be closed by orthodontic treatment to achieve desired esthetic results.[24]

Based on published reports, the overall risk of tooth getting affected or being malformed is 1 in 20 teeth, in off-springs born in consanguineous marriage between the first cousins, compared to the general population, where the risk is 1 in 40 teeth. In a way, parental consanguinity is responsible for the alteration of genotypic frequencies.[25] Consanguineous marriage increases the prevalence of rare genetic congenital anomalies and nearly doubles the risk of neonatal and childhood mortality, mental disorders, and other abnormalities in their off-springs.[26] Ruling out all the other etiological causes it was found in all the three cases that the parents were first cousins and the possibility of consanguinity was established as a probable etiological factor However, there is a scarcity of data correlating parental consanguinity with dental anomalies. Hence, Further studies are required to elucidate the possibility of their correlation.

6. Conclusion

Consanguineous marriage is a deep-rooted social trend and a respected tradition with religious beliefs in many parts of the world. Rather than discouraging consanguineous marriages in a population with such long-held tradition, ensuring pre-marital and preconception counselling would be a logical way to educate the possible risk of consanguinity with community acceptance. Increasing public literacy on consanguinity could be achieved by training primary health care workers for counselling and educating them about the prevention of congenital and genetic disorders related to consanguinity, especially in highly consanguineous populations.

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8. Conflict of Interest

There are no conflict of interest.

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