Familial Aggregation Pattern of Non-Syndromic Combined Aplasia of Maxillary Lateral Incisors and Third Molars

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ABSTRACT

Congenitally missing teeth, the most common dental anomaly, refers to teeth whose germ did not develop sufficiently to allow the differentiation of the dental tissues.

This study aimed to determine the importance of genetic investigations on oral health-related quality of life in children with congenitally missing lateral incisors.

We are presenting a genetic study performed by the A.S. Division of Medical Genetics, in Bucharest, Romania, in the case of a patient (16-year-old boy) presenting bilateral maxillary lateral incisor agenesis in the permanent dentition, a hereditary, non-syndromic familial hypodontia in three successive generations, which suggest an autosomal dominant inheritance, combined with aplasia of maxillary third molars.

The oral visual examination and the radiological exam of the patient were completed with the oral photographic examination, the family history investigation, the pedigree, and the analysis of the studied family’s genealogical tree, after acquiring the signed informed consent of the patient.

The genetic investigations of the congenitally missing lateral incisors, allow the calculation of the anomaly recurrence risk amongst offspring, and the direct and specialized monitoring of affected families, in order to limit the potential complications.

Keywords: Aplasia of maxillary lateral incisors, genetic study, hereditary, non-syndromic, pedigree.

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I. INTRODUCTION

Dental anomalies of number, known as dental agenesis, bring together the triad of anodontia, oligodontia, hypodontia, characterized by a deficit in the development of a variable number of teeth [1].

Dental agenesis, the most common dental anomaly, refers to teeth whose germ did not develop sufficiently to allow the differentiation of the dental tissues [2]-[4].

Congenitally missing teeth can affect one out of every 10 to 20 individuals worldwide and may occur, as isolated events, or within certain genetic syndromes [5]-[7].

Because the congenital absence of teeth can be isolated or syndromic, accurate diagnosis is very important for the early detection and clinical management of the patients [4], [8].

In numerous cases, where genetic investigations are not possible, determining the potential correlation between the genotype and phenotype of dental agenesis can provide valuable information leading to early and accurate medical diagnoses [8].

II. AIM

This study aimed to determine the importance of genetic investigations on oral health-related quality of life in children with congenitally missing lateral incisors.

III. MATERIAL AND METHOD

A 16-year-old man, originally from the capital region, with no reported pathological history, according to his anamnestic data, was referred to the Medical Genetics Department of A.S. Medical Center Bucharest, Romania, for a specialized genetic evaluation.

The investigation was conducted in accordance with the Declaration of Helsinki - Ethical Principles and Good Clinical Practices and after obtaining a patient's informed consent.

The case research involved the following steps: medical history, complete physical exam, extraoral and intraoral examination, radiographic examination, photographic evaluation, family history research, pedigree, analysis of the family’s tree, and laboratory tests.

IV. RESULTS

The examination of the patient face showed a mesofacial growth pattern, symmetrical and harmonious.

The clinical examination of the maxillary arch revealed the absence of both maxillary lateral incisors in the permanent dentition (Fig. 1-3).

In the lower arch, complete permanent dentition was observed, as well as diastema between the mandibular central incisors (Fig. 4).

The panoramic radiographs confirmed the bilateral agenesis of the two maxillary lateral incisors (2.1 and 2.2) associated with the absence of the maxillary third molars (1.8 and 2.8), (Fig. 5).
The patient's family history was positive in the sense that the agenesis of the maxillary lateral incisors was also highlighted in other family members, namely the proband's mother and the proband's maternal grandfather.

Based on the collected information we further performed the genealogical tree of the studied family (Fig. 6).

Genealogical tree analysis revealed hereditary, non-syndromic familial hypodontia in three successive generations (cases I:3, II:7, and III:8) suggest an autosomal dominant inheritance (I = first-generation, II = second generation, III = third generation, IV = fourth generation; □ maxillary lateral incisors agenesis; □ maxillary third molars agenesis).

V. DISCUSSION

Congenitally missing teeth, dental agenesis, congenital dental aplasia, or congenital absence of teeth, a multifactorial dental anomaly, the result of disturbances during the early stages of development, is one among the foremost common anomalies with negatively affect both esthetics and oral function through complications like malocclusion, periodontal damage, changes in jaw density, decreased chewing capacity, the difficulty of speaking, trouble eating, alterations in skeletal relationships and an unsatisfactory appearance [9].

Maxillary lateral incisor agenesis is the most common congenitally missing permanent tooth condition in the maxillary anterior region, and is the third most common developmentally absent tooth after third molars and lower second premolars, representing approximately 20% of all dental anomalies [10], [11]. The missing upper lateral incisor has been found to be more prevalent in females and bilateral cases are more frequently reported than unilateral [10].

Largely, dental agenesis has been attributed to genetic factors, but they may also be caused by environmental factors [10]. Additionally, can occur in a number of conditions such as ectodermal dysplasia, cleft lip and palate, Down's Syndrome, Incontinentia pigmenti and following early irradiation [11].

The susceptibility of maxillary lateral incisors to dental agenesis has been associated with their anatomical position in the maxillary arch and also the fact that they are the last teeth to develop in their respective classes [10].

Congenitally missing lateral incisors whether unilateral or bilateral may affect the self-esteem and social relationships of the affected individual, may cause a variety of esthetic and functional problems [12], [13]. Their absence may cause a diastema between the central incisors, spacing between permanent incisor and canine, mesial migration of canines, and midline shift in case of a unilateral missing tooth [13].

Besides an unfavorable appearance, patients with missing teeth may suffer from malocclusion, periodontal damage, insufficient alveolar bone growth, reduced chewing ability, inarticulate pronunciation and other problems [14]-[18].

Moreover, the treatment of congenitally missing lateral incisors is a complex and challenging process, which demands the interaction of several dental specialties [13], [19]-[22].

VI. CONCLUSION

The case report, illustrates a bilateral absence of the maxillary lateral incisors present in the permanent dentition, a hereditary, non-syndromic familial hypodontia in three successive generations (cases I:3, II:5, and III:9), which suggest an autosomal dominant inheritance. The genetic investigations of the congenitally missing lateral incisors, allow the calculation of the anomaly recurrence risk amongst offspring, and the direct and specialized monitoring of affected families, in order to limit the potential complications.

AUTHORS' CONTRIBUTION

All authors contributed equally with the first author, in the preparing, review, and editing of the article.

All authors read and approved the final version of the manuscript.

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CONFLICT OF INTEREST

Authors declare that they do not have any conflict of interest.

STATEMENT OF INFORMED CONSENT

Informed consent was obtained from all individual participants included in the study.

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