ejpmr, 2021,8(2), 85-88

SJIF Impact Factor 6.222

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

Case Study

ISSN 2394-3211

EJPMR

NONSYNDROMIC FAMILIAL HYPODONTIA: GENETIC STUDY

Cristina-Crenguta Albu*, Marina Imre, Ana Maria Cristina Tancu and Stefan-Dimitrie Albu

University of Medicine and Pharmacy "Carol Davila", 37 Dionisie Lupu Street, 1st District, 020021, Bucharest, Romania.

***Corresponding Author: Cristina-Crenguta Albu**

University of Medicine and Pharmacy "Carol Davila", 37 Dionisie Lupu Street, 1st District, 020021, Bucharest, Romania.

Article Received on 10/12/2020 **Article Revised on 30/12/2020** **Article Accepted on 20/01/2021**

ABSTRACT

Hypodontia is a dental number anomaly characterized by the partial absence of teeth. We are presenting the family genetic study of hereditary, nonsydromic hypodontia, illustrating the heterogeneity of phenotypic expression of hypodontia, amongst two successive generations. The genetic study was performed by the Genetics Department of A.S. Medical Center, in Bucharest, Romania. The clinical and paraclinical examination of the patients was completed by the photographic examination, the family investigation, the genealogical tree and the analysis of the studied family's genealogical tree, after acquiring the informed consent of the patients. The genetic study allows for the assessment of the dental number anomalies, from the point of view of the genetic factor involvement in their etiology, it facilitates determining the disorder's recurrence risk within the studied family, it allows for the early diagnosis of the disorder, preventing complications.

KEYWORDS: Nonsyndromic, hypodontia, genetic study, autosomal dominant, family tree.

INTRODUCTION

Hypodontia is a dental number anomaly, characterized by the partial absence of teeth.[1-3]

It affects approximately 7% of the European population and mainly the female sex.[4] The absence of the last molar was not included in this estimation.

In the European population, the second, inferior premolar, the lateral superior incisors and the second superior premolar are more frequently missing.[5-7]

Hypodontia may occur as an isolated event or in association with various system disorders.[8]

MATERIALS AND METHODS

We are presenting the case of a family: mother and son, who checked into the A.S. Medical Center, from Bucharest, Romania, for a genetic examination.

The genetic study was performed by the Genetics Department of our medical center, and it entailed the following stages: the anamnesis, the local and general medical examination, the radiological examination, the photographic examination, the family investigation, the genealogic tree and the analysis of the studied family's genealogical tree, after having acquired their consent.

RESULTS

The mother, case II4 of the family tree, a 42-year-old patient from the urban area, with a normal phenotypic expression and without associated syndromes, presented at the oral clinical examination, agenesis of both mandibular lateral incisors 3.2 and 4.2 (Fig. 1).

The son, case III6 of the family tree, aged 6, also displays normal phenotypic expression and has no associated syndromes. At the oral clinical examination, the agenesis of the mandibular central incisor 3.1, was revealed (Fig. 2).

The radiological examination, which was performed with great difficulty due to difficult cooperation, revealed the additional absence of the 1.7, 2.7 and 4.7 tooth buds (Fig. 3).

Based on the data collected and recorded in the medical file for congenital malformations, we further performed the genealogical tree of the studied family (Fig. 4).

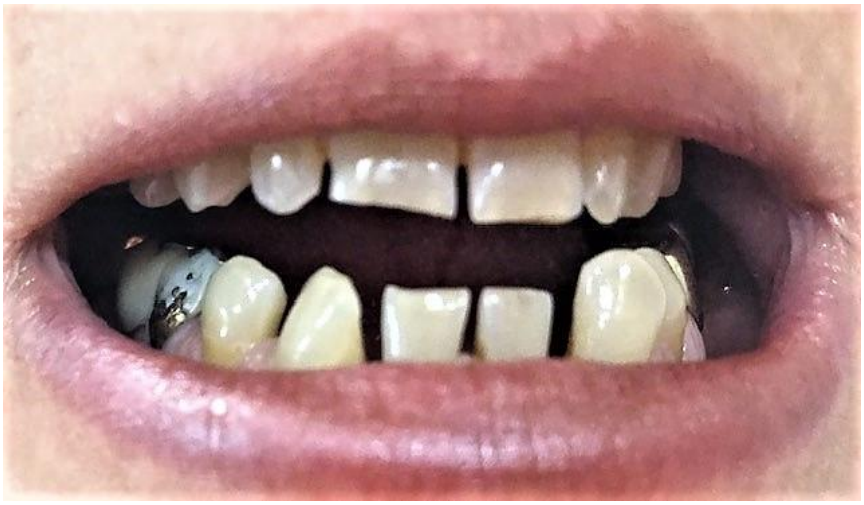

Figure 1: Oral clinical examination of the case II4 of the family tree (the mother) highlights the agenesis of the mandibular lateral incisors 3.2 and 4.2.

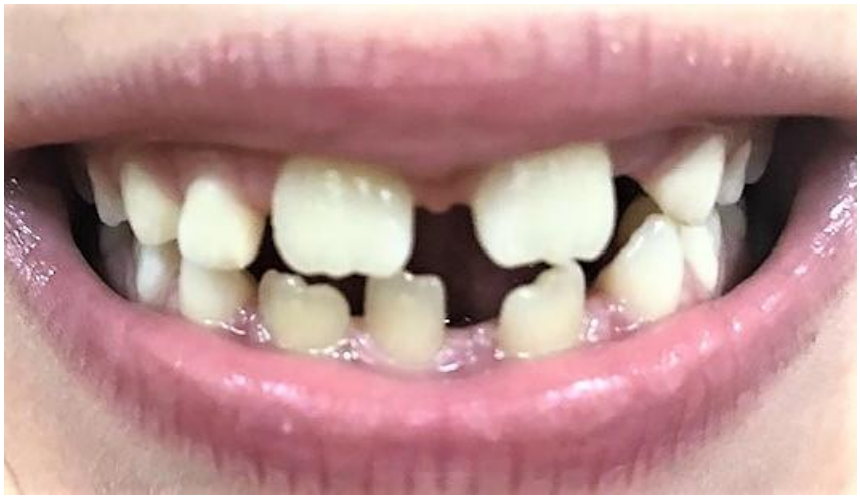

Figure 2: Oral clinical examination of the case III6 of the family tree (the son) highlights the agenesis of the mandibular central incisor 3.1.

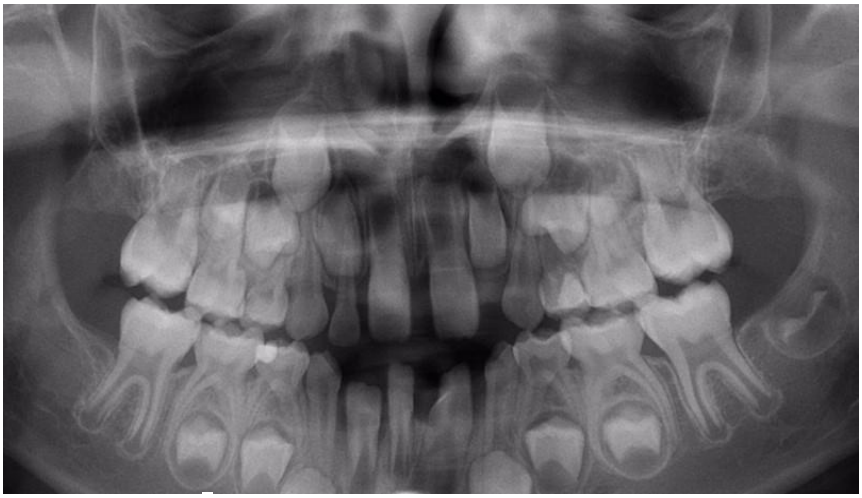

Figure 3: Radiological examination of the case III6 of the family tree (the son) highlights the agenesis of the mandibular central incisor 3.1, and the absence of the 1.7, 2.7 and 4.7 tooth buds.

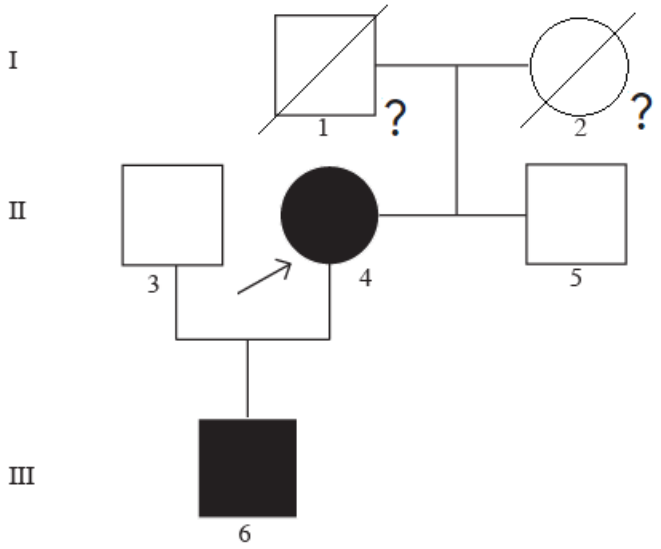

Figure 4: Genealogical family tree: I = first generation, II = second generation, III = third generation. The case II4 of the family tree highlights the mother, and the case III6 of the family tree highlights the son.

The analysis of the genealogical tree illustrates a non-syndromic familial case of hypodontia in the permanent dentition, with autosomal dominant inheritance, present in two successive generations, with variability of phenotypic expression.

DISCUSSION

As an isolated dental anomaly, hypodontia is considered a hereditary condition.[9,10] It is controlled by a mutant allele, autosomal dominant with incomplete penetrance and variable expressivity.[11-13]

As associated dental anomaly, hypodontia occurs in different genetic syndromes, such as ectodermal dysplasia, the Ellis van Creveld syndrome, the Book syndrome, incontinentia pigmenti (the Bloch-Sulzberger syndrome).[14-17]

CONCLUSION

The presented case illustrates a familial case of hypodontia, clinically manifested in the permanent dentition, with autosomal dominant inheritance, highlighted in two successive generations and characterized by phenotypic expression variability.

The genetic study allows the assessment and evaluation of dental number anomalies, from the point of view of the implication of the genetic factor in their etiology, facilitates the determination of the recurrence risk of the condition within the studied family, allows early diagnosis of these abnormalities, personalized monitoring of family cases, as well as preventing complications.

Authors' contributions

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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