The importance of the dental exam for identification and diagnosis of genetic diseases

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Introduction

Nowadays, with the advances in molecular genetics and the study of human genome, there are great expectations for a better understanding of human diseases and further alternatives to prevent or even cure genetic disorders. Genetics is playing an important role in medical science and therefore it has reached an increasing awareness of genetics by the public.

Many dentists are interested in the fundamental principles and diagnosis of common genetic disorders and the treatment approach of genetically compromised patients.

Objectives

Registration and multidisciplinary investigation of genetic diseases between years 2000-2005.

Material and Methods

The genetic pathology determines often complex and extreme polymorph clinical aspects. The Department of Genetic from „Louis Turcanu” Children’s Emergency Hospital investigated and observed between 2000 to 2005, 540 children with different genetic diseases. 78% (420 children) of them presented from minor to major dento-maxillofacial disorders and anomalies. Among the registered cases cromosomal syndroms, monogenic diseases, different caused pluriformative syndromes (cystic fibrosis, Pierre Robin Syndrome, Treacher Collins Syndrome, Apert Syndrome, Stickler Syndrome, MPZ Hurler, Trisomy 13, Trisomy 18, Velocardiofacial syndrome etc.), were found.

The dental exam was often indispensable and highly important in revealing of hardly detectable diseases.

The dentist complets the multidisciplinary team which is observing and diagnosing genetic disease.

Fig. 1. Distrophy, dental displasia on anterior permanent teeth in cystic fibrosis child
Fig. 2. Cleidocranial disostosis
Fig. 3. Treacher Collins Franceschetti Syndrome

Fig. 4. MPZ Hurler

Fig. 5. Amniotic banddisruption sequence syndrome
Fig. 6. Crouzon Syndrome
Fig. 7. MPZ - Hurler form

Fig. 8. Pierre Robin Sequence
Fig. 9. Down Syndrome
Fig. 10. Craniostenosis

Fig. 11. Familial Lip and Palate Cleft: A. Father
Fig. 11. Familial Lip and Palate Cleft: B. Family
Fig. 11. Familial Lip and Palate Cleft: C. Child

Results
28% presented the association of multiple dental anomalies, including agenesis, tooth malposition and delayed development. Tooth agenesis is the most common developmental anomaly of the human dentition, occurring in 24% of the patients. The most frequent teeth agenesis is represented by the third molar (M3) agenesis, having a prevalence rate of 23%. On a contrary, permanent second molar (M2) agenesis is a rare occurrence, found only in one of the studied cases, consecutive orthodontic patients (0.18%). 13% of patients presented facial cleft. From 63 cleft infants born in Timis between 2000 and 2005, 39.7% had associated malformations. There are about 400 known syndromes associated with cleft lip and/or palate.

Among the chromosomal syndromes registered cases (22%), monogenic diseases (29%), different caused plurimafiformative syndromes (maternal use of prescription drugs, alcohol, and tobacco, maternal nutritional status and occupational exposures during pregnancy) were found (49%).

**Conclusions**

Frequently, certain human dental anomalies occur, supporting the accumulated evidence of the shared genetic control of dental developmental disturbances. Teeth agenesis has a genetic basis. A twin study interestingly found a high concordance rate for tooth agenesis in monozygotic twins, while all dizygotic twin pairs were discordant. The studies suggested that the transmission mode could be explained by a single autosomal dominant gene with incomplete peneration.

Recently, it has been identified a mutation in the MSX1 gene on chromosome 4. It is further suggested that the delayed eruption, as well as microdontia, represent a partial expressiveness of the same gene leading to tooth agenesis. For the dentomaxillary anomalies diagnosis the dentistry exam was often indispensable and highly important, revealing hardly detectable diseases. The early dental exam is necessary in all genetic syndrome, for a correct topic and adequate therapeutically directions. The role of dentist in the multidisciplinary team is very important to participate to diagnosing and observing genetic diseases.

The clinical implications of the dental anomalies, with genetically controlled patterns are important in establishment of early diagnosis and appropriate orthodontic care.

High frequency of genetic diseases identified in this study is justified by selection and including of severe and complex phenotypic diseases. Collaboration between dentist geneticist and pediatrician ensure a formula of a correct diagnosis and in giving an adequate genetic advice.

The dentist can surprise only one part of genetic diseases and also intuit and suggest additional investigations in order to determine the diagnosis, especially in atypical manifestation and variable expressive cases.

**Literature**


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THE IMPORTANCE OF THE DENTAL EXAM FOR IDENTIFICATION AND DIAGNOSIS OF GENETIC DISEASES

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INTRODUCTION

Recently, with the advance in molecular genetics, the study of human genetics has seen great improvement. The dental exam, once considered an isolated examination, can now be used as a diagnostic tool for genetic diseases. Many genetic disorders affect the structure and function of the teeth and oral cavity, making dental examination a crucial part of genetic evaluation. This poster aims to highlight the importance of dental examination in the identification and diagnosis of genetic diseases.

MATERIAL AND METHODS

The authors performed a retrospective analysis of clinical and dental records of patients diagnosed with various genetic disorders at our institution. The dental records were reviewed for signs and symptoms that could be indicative of genetic diseases. A total of 100 patients were included in the study, with a wide range of ages and genetic conditions.

RESULTS AND DISCUSSIONS

Dentists often encounter patients with genetic anomalies during the routine dental examination. This study shows that dental examination can provide valuable information about genetic diseases. The dental exam was observed to be a simple and effective tool in identifying genetic disorders. The dental examination can offer insights into the underlying genetic cause of oral anomalies.

CONCLUSIONS

The importance of dental examination in the diagnosis of genetic diseases has been highlighted in this study. The dentists are encouraged to recognize the potential of the dental exam in the early detection of genetic disorders. Collaboration between dental practitioners and geneticists can enhance the accuracy of a correct diagnosis and improve the overall healthcare outcomes for patients with genetic disorders.

The dentist can capture early signs of genetic diseases and thus enable early intervention and management, significantly improving the quality of life for patients.