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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 therfore 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 determins often complex and extreme polimorph 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 registrated cases cromosomial syndroms, monogenic diseases, different caused plurimalformative 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 diagnosting genetic disease.

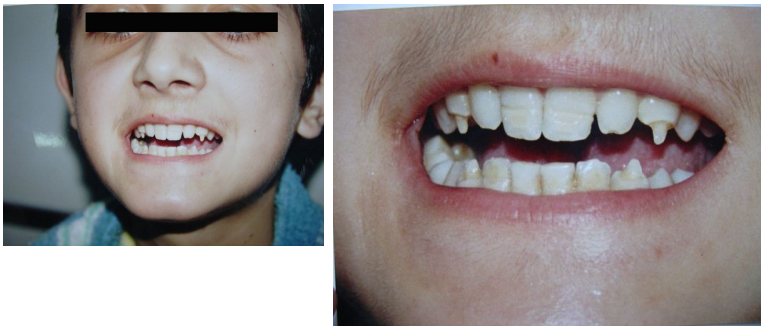


Fig. 1. Distrophy, dental displasia on anterior permanent teeth in cystic fibrosis child

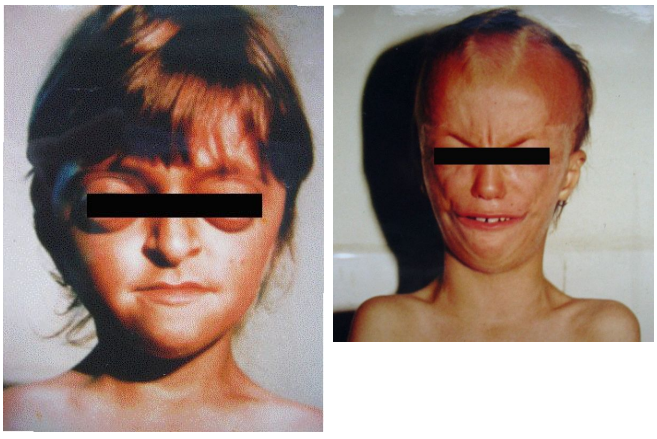


Fig. 2. Cleidocranian disostosis

Fig. 3. Treacher Collins Franceschetti Syndrome

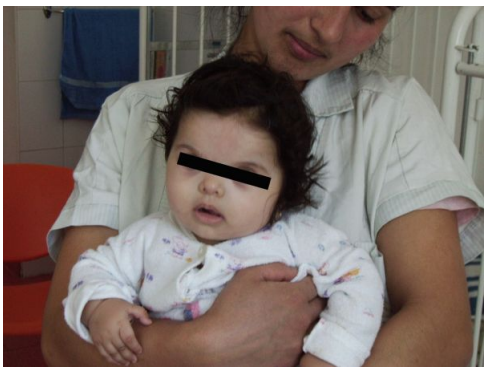


Fig. 4. MPZ Hurler

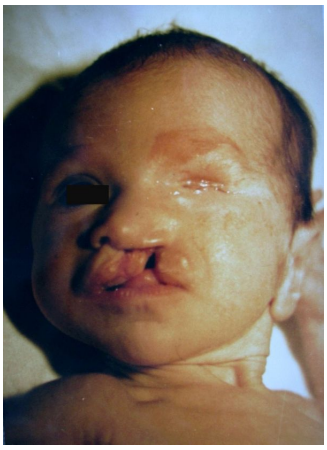


Fig. 5. Amniotic banddisruption sequence syndrome

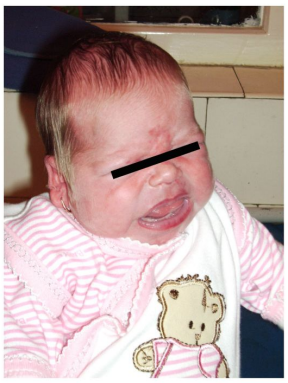


Fig. 6. Crouzon Syndrom

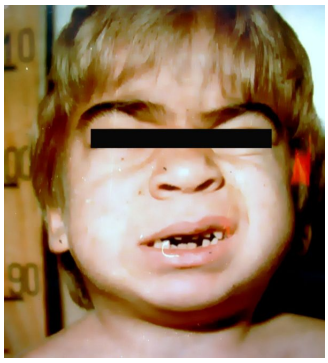


Fig. 7. MPZ - Hurler form

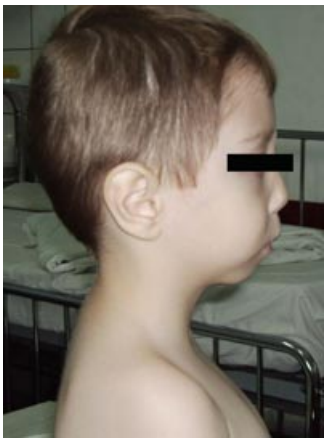


Fig. 8. Pierre Robin Sequence

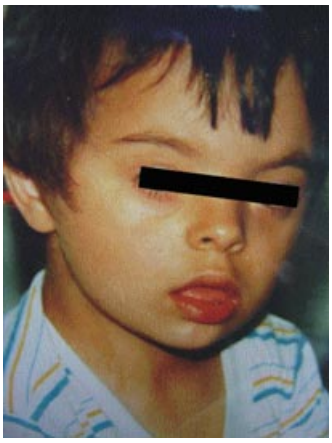


Fig. 9. Down Syndrome

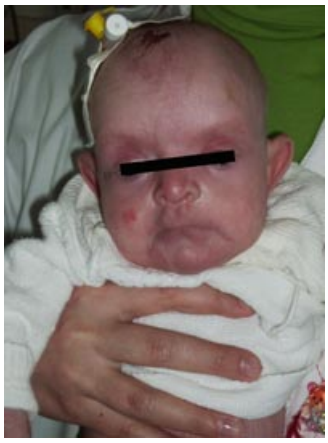


Fig. 10. Craniostenosis

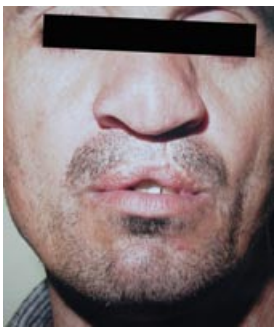


Fig. 11. Familial Lip and Palate Cleft: A. Father

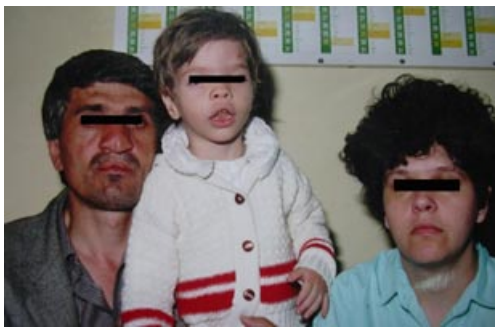


Fig. 11. Familial Lip and Palate Cleft: B. Family

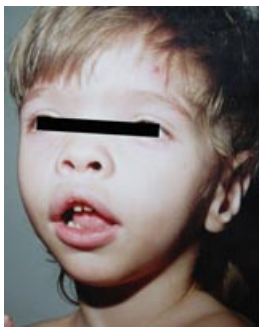
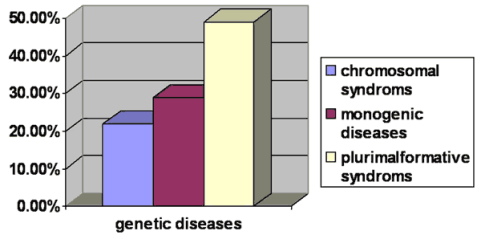


Fig. 11. Familial Lip and Palate Cleft: C. Child

Results

28% presented the association of multiple dental anomalies, including agenesis, tooth mallposition 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 registrated cases (22%), monogenic diseases (29%), different caused plurimalformative 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 singtle 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 multi disciplinary 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

1. Chaventre A., Cazes M.H., Roth A., Genetique des populations de isolats. Flamarion Medicine, 1981.
2. Cohen J.S., Hogman M.E., The new genetic medicines, Scientific American, 1994, december, 77-82
3. Emery A.H., Rimoin D.L., Principles and Practice of Medical Genetics, Churchill Livingston, vol. 1., 1983
4. Newton Freire M.,Effects of consanguineous Marriages on Morbidity and Precocious Mortality Genetic Counseling. Am.J. Med. Genet,18, 401-405, 1984
5. Vogel F., Motulsky A.G., Human Genetics, Springer Verlag, Berlin-Heidelberg, 1986
6. Peters H, Balling R, Teeth. Where and how to make them. Trends Genet. 1999, 15: 59-65
7. Mensah J.K.,Ogawa T.,Kapadia H., Cavender A.C., D\'Souza R.N., Functional analysis of mutation in PAX9 associated with familial tooth agenesis in humans. J.Biol. Chem.2004, 279:5924-5933.

This Poster was submitted by Prof. Cristina Bortun.

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Poster Faksimile:

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

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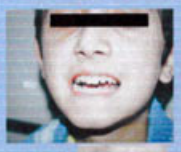
"VICTOR BABEŞ" UNIVERSITY OF MEDICINE AND PHARMACY
FACULTY OF DENTISTRY-SPECIALIZATION OF DENTAL TECHNOLOGY



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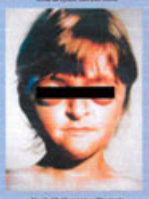

Fig. 2. Childsyndrome illustrate

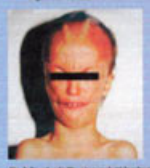

Fig. 3. Treacher Collins / Franceschetti Syndrome

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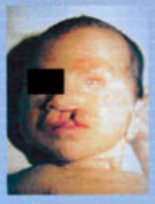

Fig. 5. Anxiety bandibreption sequence syndrome

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 therfore 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.

MATERIAL AND METHODS

The genetic pathology determins often complex and extreme polimorph 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. 70% (420 children) of them presented from minor to major dento-maxillofacial disorders and anomalies. Among the registrated cases craniosostal syndroms, monogenic diseases, different caused pluricausalitetmotive syndromes (cystic fibrosis, Pierre Robin Syndrome, Treacher Collins Syndrome, Apert Syndrome, Stickler Syndrome, MPZ Harlot, 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 completes the multidisciplinary team which is observing and diagnosting genetic disease.

RESULTS AND DISCUSSIONS

Frequently, certain human dental anomalies occur, supporting the accumulated evidence of the shared genetic control of dental developmental disturbances. 20% 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. (orthodontic patients (0. 18%). Teeth agenesis has a genetic basis. A twin study interestingly found a high concordance rate for teeth 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 penetration.
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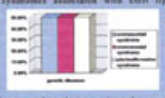

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CONCLUSIONS

For the dentomaxillary anomalies diagnosis the dentistry exam was often indispensible and highly important, revealing hardly detectable diseases. The early dental exam is necessary in all genetic syndroms, for a correct topic and adequate therapeutically directions. The role of dentist in the multi disciplinary team is very important to participate to diagnosing and observing genetic diseases.

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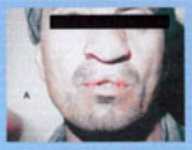
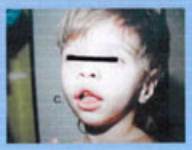


Fig. 11. Fissilial Lip and Palate Cleft: A. Father, B. Family, C. Child



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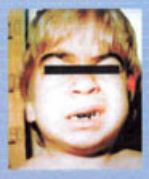

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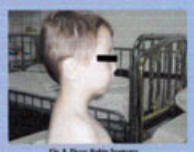

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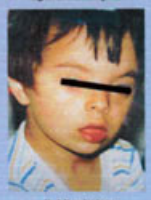

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