Facial and dento-maxillary expressions in genetic discorders

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Summary

The congenital malformations with craniofacial expressions are on the fourth place in the human pathology and they are frequently associated with other anomalies.

Our study refers to some genetic disorders, that are accompanied by facial and dento-maxiilary expressions. Therefore we selected certain syndroms, such as Crouzon, Treacher Collins, Hurler, cleft lip and palate, that allow a dentist to recognize a genic disorder.

Whenever a dentist suspects a genetic disorder, he should ask for cooperation with a medical geneticist, to formulate an exact genetic diagnosis.

Key words: craniofacial malformations, genetic disorders, facial and dento-maxillary expressions, cooperation dentist - medical geneticist.

Introduction

The new concept in medicine requires knowledge of the relationship between dento-maxillary disorders and medical genetics.

In fact, any disease is the result of the combined action of the genetic component and the environment, but in many disorders, the role of the genetic component is wider, even complete. Knowledge of the dento-maxillary disorders, associated with a genetic disease is very important [1,2, 3].

In medical practice any genetic disorder has dento-maxillary expressions. Therefore, whenever a dentist suspects a genetic disorder, he should ask for cooperation with a medical geneticist, to formulate an exact and fast genetic diagnosis.

Among disorders caused completely or partly by genetic factors, three main types are recognized:

- single gene disorders;
- chromosome disorders; polygenic
- multifactorial disorders.

Of all genetic disorders accompanied by facial and dento-maxillary expressions, we selected certain syndroms with evident signs, like Hurler, Crouzon, Treacher Collins, mucovis-cidosis, cleft lip and palate, that allow a dentist to recognize a genetic disorder.

The mucoviscidosis is a hereditary disease that is present from birth. It is caused by a defect in a recessive type gene. The cystic fibrosis causes certain glands in the body to produce abnormal secretions that can cause many symptoms. The most important of these symptoms affect the digestive tract and the lungs. Throughout the world approximately one person in every 22 is a carrier even though the chance of two carriers reproducing is around 500 to one [3, 4].

The **mucopolysaccaridoses** are a heterogenous group of storage diseases in which mucopolysaccarides accumulate in lysosomes as a result of a deficiency of one of the enzymes required for their degradation. The most severe is the **Hurler syndrome.** Affected children are mentally retarded, have skeletal abnormalities, short stature and other abnormalities [1, 3,4].

The Crouzon syndrome is characterized by craniosynostosis (acrocephaly, brachycephaly, flat occiput, palpable ridging, high prominent forehead +/- frontal bossing) and dysmorphic facial features (flattened face with maxillary hypoplasia, relative mandibular prog-nathism, ears with low set and conductive hearing loss, ocular features, short upper lip +/- cleft lip, class **III** malocclusion with maxillary crowding, high-arched narrow palate +/- cleft palate, bifid uvula). It is caused by an autosomal dominant gene, located on the 10q25-26 chromosome. The incidence is 1/25,000 [1,3].

Treacher Collins Franceschetti syndrome is a disorder of craniofacial development. The features include antimongoloid

slant of the eyes, coloboma of the lid, micrognathia, microtia and other deformity of the ears, hypoplastic zygomatic arches, and macrostomia. Conductive hearing loss and cleft palate are often present. Inocclusions or malocclusions are also often present. Treacher Collins syndrome is an autosomal dominant disorder; the gene is located on the 5q31.3-32 chromosome [1, 3].

Cleft lip with or without **cleft palate** is one of the most common congenital malformations caused by a failure of fusion of the frontal process with the maxillary process at about 35th day of gestation. About 60-80 percent of those affected are male.

Cleft lip and cleft palate are heterogenous and include isolated single gene forms, numerous single gene syndromes, polygenic multifactorial threshold trait, forms associated with chromosomal disorders and many cases resulting from teratogenic exposure [3, 5, 6, 7].

Materials and methods

A number of 56 patients (46 children and 10 adults) were consulted in genetic counseling, dental and pediatric praxis.

We used two types of investigations:

- specific: genetic and dental investigations;
- unspecific: for the specification of the exact genetic diagnosis and the severity of the disease.

Results and discussions

The distribution of the cases, dependent on the etiology, are shown in *Table 1*.

Table 1. Distribution of the cases dependent on the etiology

Etiology		Number of patients	Percent (%)
genetic	single genic	20	35.5
	chromosomial	12	21.5
polygenic multifactorial		12	21.5
epigenetic		12	21.5

The most common genetic disorders associated with facial and dento-maxillary expressions in our patients were:

- mucoviscidosis;
- Hurler syndrome;
- Crouzon syndrome;
- cleft lip and cleft palate;
- Treacher Collins Franceschetti syndrome.

Figures 1-4 show some significant cases of genetic disorders, with craniofacial and dental changes.

As any genetic disorder has dento-maxillary expressions, the dentist can be the first that suspects a genetic disorder. Therefore, he should ask for another investigation and a cooperation with a medical geneticist because the role of the genetic component is wider [7].

Figure 1. Dental distrophy in mucoviscidosis

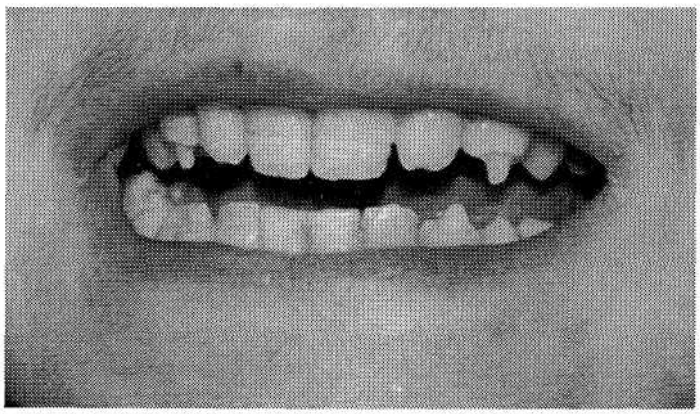


Figure 2. Craniofacial changes in Crouzon syndrome

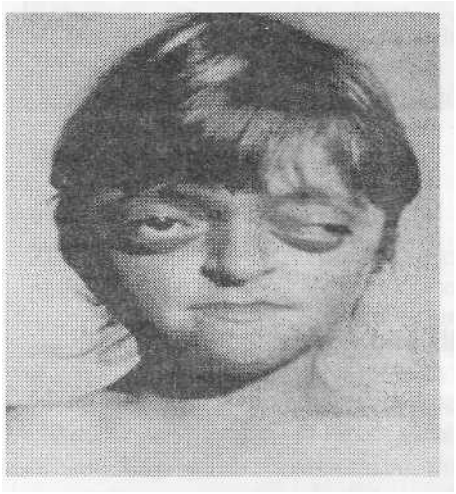


Figure 3. Cleft lip and cleft palate

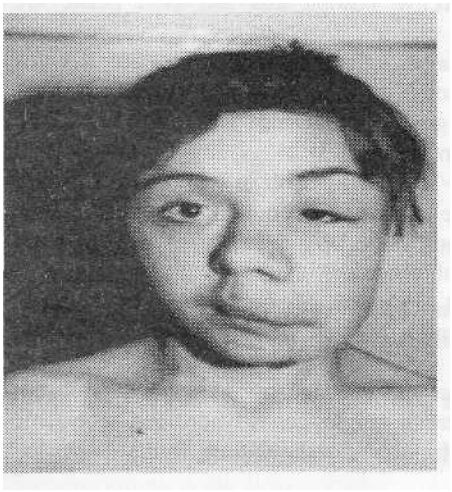
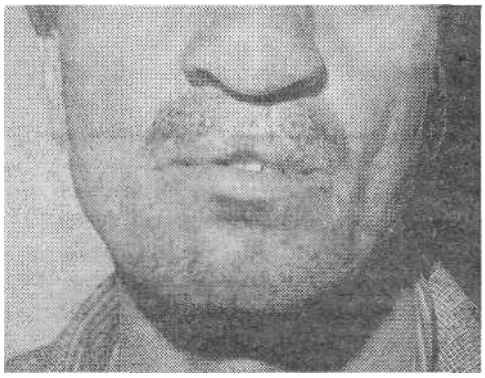
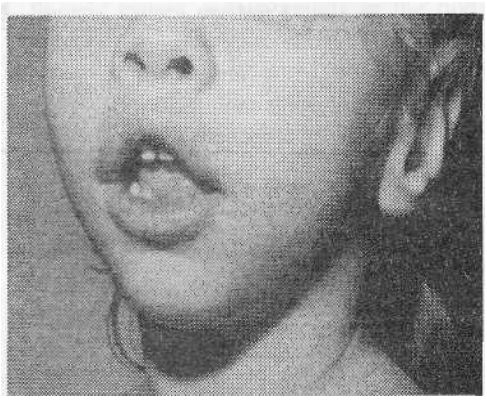


Figure 4. Craniofacial changes in Treacher Collins Franceschetti syndrome





Conclusions

1. The great frequency of the genetic disorders in our patients is legitimated by the selection and including of cases with great affectation of the phenotype.

2. The dentist can be the first who consults the patient and he should ask for other investigations when he suspects a genetic disorder;

3. A cooperation dentist - medical geneticist - pediatrician leads to settling an exact genetic diagnosis.

References

1. Bortun C, Puiu M., Leretter M., Sandu L. Modificari faciale si dento-maxilare in bolile genetice, Congres international - Sanatatea orala in tarile bazinului Marii Negre, Constanta, 21-24 septembrie 2002. 2. Puiu M., Tudose O., Bortun C. Modificari dento-maxilare si faciale in sindroamele

cromozomiale, Congres international - Sanatatea orala in tarile bazinului Marii Negre, Constanta, 21 -24 septembrie 2002.
3. Tudose Olimpia. Genetica medicala, Lito UMF Timisoara, 1996.
4. Puiu Maria. Mic dictionar de genetica medicala, Ed. Eurobit, Timisoara, 1998.
5. Grivu O., Cristoloveanu R., Mecher E. Stomatologie pediatrics, Ed. didactica s.i pedagogica, Bucuresti, 1975.
6. Knecht A.K., Bronner-Fraser M. Induction of the neural crest: a multigene process. *Nature Reviews Genetics*, 2002, 3: 453-461.
7. Wilkie A.O.M., Morriss-Kay G.M. Genetics of craniofacial development and malformation. *Nature Reviews Genetics* 2001; 2: 458-468.

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