Dentomaxillary abnormalities in genetic diseases

Gabriela Lilios[1], Lenuta Malcea[2]

Constanta, Romania

Summary

The purpose of this work is to identify the congenital bone diseases (as a result of the anomalies in the bone and joint system development), associated with dentomaxillary modifications.
Objectives. **Stating the etiology of congenital bone diseases, in order to anticipate dental problems, their prognosis and recurrence risk. Approaching various types of dental anomalies, their severity and degree of spreading in the most frequent congenital bone diseases. Identifying those bone syndromes, which benefit from etiopathogenic, general, orthopedic, surgical etc. treatment supporting the dental recovery effort.**
Methods. **The survey is a five-angled approach of the most frequent congenital bone diseases related to dentomaxillary anomalies, bone general signs of the considered syndromes; etiology; type of related dentomaxillary modifications.**
Results**. Genetic bone diseases are associated with dentomaxillary modifications, as common alterations occur ever since the embryonic stage. Enzymopathies, which result in the unmetabolized substances storage, at bone level included, alter facial bones and dentition, as well. Determination of the type of bone disease, as well as the etiologic classification is necessary to anticipate dental mishaps, in order to determine their prognosis and recurrence risk.**
Conclusions**. Disorders in the dentition development take on prevailing aspects in case of a certain bone disease, however they may co-exist. Etiopathogenic treatment improves the overall evolution of such diseases, supporting the dental recovery effort.**

Key words**: dentomaxillary anomalies, embryological development, bone, new-born.**

Introduction

The survey approaches the issues on dentomaxillary modifications, their types, complexity and spreading degree occurring in the bone and joint system development anomalies. The purpose of our endeavor is to identify the bone syndromes accompanied by dentomaxillary modifications, assuming that they have the same embryologic origin (*Table 1*), as alterations in the embryologic stage entail dentomaxillary modifications.

Material and Method

This work approaches the main bone signs of malformation syndromes (as secondary to the anomalies in the bone and joint system development) and coexisting dentomaxillary abnormalities, in descriptive manner, as from the simultaneous and com-

[1] Assoc. Prof., M.D., Department of Physiology, Faculty of Dental Medicine and Pharmacy, "Ovidius" University of Constanta
[2] M.D., Pediatrician and Neonathologist, Clinical Hospital of Constanta

Table 1. Embrionic development. Face and skeleton. (Jones, 1988)

Age	**Face**	**Skeleton**
24 days	Mandible Hyoid arches	
30 days	Fusion, mandible, arches	Arm bud
34 days		Leg bud
Weeks	**Fetal development**	
7 1/2	Palatal swellings Dental lamina Epithelium	Cartilaginous models of bones Chondrocranium Tail regression
8	Nares plugged Sublingual gland	Ossification center
10	Lips Palate	Sternum Joints
12	Dental papilla Notochord degenerated.	Tail degenerated
16	Palate complete Enamel and dentine	Distinct bones
24	Nares reopen Calcification of tooth primordial	
38	Rudimentary frontal maxillary sinuses	

mon embryologic development of the skeleton and facial bones.

The following are considered:

1. congenital bone diseases frequently triggering dental abnormalities, as well as those where such alterations occur occasionally;

2. more frequent types of dental modifications accompanying such syndromes;

3. their etiology and transmission ways in order to determine their prognosis and recurrence risk;

4. possible related mental deficit;

5. etiopathogenic, general, orthopedic, and surgical treatment.

Results

I. Anodontia

Frequent in:

1. Albright Hereditary Osteodystrophy Syndrome Pseudohypoparathyroidism – because of hypocalcemia and hyperphosphatemia that are unresponsive to parathormone [1]. *Dentition*: Delayed dental eruption, aplasia and/or enamel hypoplasia.

Etiology: the findings of a 2:1 female to male sex incidence have favored an X-linked dominant mode of determination.

Treatment: Vitamin D therapy in a dosage of 25,000 to 100,000 units per day may be necessary. The therapy should be discontinued every few years, because spontaneous amelioration of the hypocalcemia may occur with time.

2. Chondroectodermal Dysplasia

*Dentition***:** neonatal teeth; partial anodontia; small teeth, and/or delayed eruption; dental problems are frequent.

Etiology: Autosomal recessive.

The majority of survivors are of normal intelligence.

3. Cleidocranial Dysostosis, a "constitutional", generalized and symmetric disease. The more obvious feature is the defect in clavicle and cranium [2].

Abnormalities: craniofacial (brachycephaly with bossing; midfacial hypoplasia); clavicle and chest (aplasia of clavicle; small thorax).

Dentition: late eruption (especially the permanent teeth).

Malformed roots; retention cysts; enamel hypoplasia; supernumerary teeth.

Etiology: autosomal dominant, with wide variability in expression.

The patients have a normal intelligence.

4. Frontometaphyseal Dysplasia

*Abnormalities***:** craniofacial (coarse face with prominent supraorbital ridges). Limbs (flexion defects of fingers, wrists, elbows, knees, and ankles).

Chest deformities, scoliosis.

*Dentition***:** partial anodontia; delayed eruption; retained deciduous teeth; high palate; small mandible.

*Etiology***:** X-linked with severe manifestations in male and variable but more mildly affected females.

5. Oto- palato digital Syndrome (Taybi Syndrome)

The syndrome was initially described by Taybi in 1962. After that many cases have been recognized.

Abnormalities: small stature; cranium (frontal and occipital prominence; absence of frontal and sphenoid sinuses); facial bone hypoplasia; limb (limited elbow extention).

*Dentition***:** partial anodontia; impacted teeth.

Performance: mild mental deficiency (IQ = 75-90).

Etiology: X-linked, semidominant.

6. Pycnodysostosis

In 1962, Maroteaux and Lamy described the special condition (cleidocranial dysostosis associated with osteosclerosis and bone fragility) as pycnosynostosis.

Abnormalities: small stature; osteosclerosis; craniofacial (frontal and occipital prominence; delayed closure of sutures; facial hypoplasia).

Dentition: irregular permanent teeth; partial anodontia; delayed eruption; caries.

Etiology: autosomal recessive determination.

The artist Toulouse Lautrec is considered to have had pycnodysostosis.

Occasional in:

1. Crouzon Syndrome

*Abnormalities***:** shallow orbits; premature craniosynostosis; maxyllary hypoplasia.

Dentition is affected through maxillary hypoplasia.

Etiology: Autosomal dominant with variable in expression. About one quarter of the reported cases have had a negative family history.

2. Oculo-dento digital Syndrome

Abnormalities: Eyes (microphtalmos, microcornea, short palpebral fissures).

Dentition: enamel hypoplasia.

Etiology: dominant autosomal determination.

II. Hypodontia

Frequent in:

1. Chondroectodermal dysplasia

2. Coffin Lowry Syndrome

Dentition: hypodontia; malocclusion; wide spaced teeth.

Etiology: X-linked determination. The females have slight to moderate mental deficiency.

3. Down Syndrome (trisomy 21 syndrome).

This syndrome has an incidence of 1/660 newborns making it the most common pattern of malformation in man.

Abnormalities: general, CNS, craniofacial, ears, neck, hands, feet, pelvis, cardiac etc.

Dentition: hypoplasia, irregular placement, fewer caries than usual.

Performance. Mental deficiency (IQ =

25-50 with an occasional individual above 50, the mean IQ for older patients is 24).

Etiology. Trisomia 21.

Types of chromosomal alteration of Down syndrome: full 21 Trisomy - 94 %; 21 Trisomy/normal mosaicism – 2.4 %; translocations cases – 3.3 %; Mosaicism usually leads to a less sever phenotype.

4. Osteogenesis imperfecta, type I

At least four type of osteogenesis imperfecta exist. Osteogenesis imperfecta associates dental disorders [3].

Abnormalities: small stature; bones (thin cortices, small facial bones); joints and ligaments; sclerae and skin; hearing.

Dentition: hypoplasia of dentin and pulp with translucency of teeth; yellowish or bluish-gray coloration; susceptibility to caries; irregular placement; late eruption.

Natural History. There is wide individual variability. The long leg bones are the most frequent sites of breakage. After adolescence, the likelihood of fractures diminishes. By 30-39 years of age, 35% of patients are deaf; by 60 years of age 50% of patients are deaf.

*Etiology***.** Autosomal dominant.

*Treatment***.** Long term calcitonin may be beneficial in reducing the frequency of fracture. Also, fluoride by yielding a stronger calcium apatite crystal may be beneficial.

5. Pycnodisostosis

6. Tricho-Dento-Osseous Syndrome

Abnormalities: kinky hair at birth; facies with frontal bossing, dolicocephaly; bones with mild to moderate increased density.

Dentition: small, widely spaced; pitted teeth with poor enamel; increased pulp chamber size (taurodontism).

The teeth become eroded and discolored, are prone to periapical abscesses and/or lost by the second to third decade.

Etilogy: autosomal dominant.

III. Enamel hypoplasia

Frequent in:

1. Morquio Syndrome (Mucopolysaccharidosis IV).

Deficiencies of two different enzymes leading to a severe form: mucopolysaccharidosis IV A, and a mild form, mucopolysaccharidosis IV B.

Abnormalities: severe limitation of growth (adult stature 82 to 115 cm); craniofacial; skeletal and joints; marked platyspondily; short, curved long bones, short stubby hands.

Dentition: widely spaced teeth with thin enamel that tends to become grayish in color.

Mentality is normal in both the severe and the mild forms.

Etiology: autosomal recessive.

2. Pseudovitamin D Deficiency Rickets

There is a disease described by Prader in 1961.

Metabolic: hypocalcemia, elevated serum alkaline phosphatase.

Dentition: enamel hypoplasia of postnatal onset.

*Treatment***.** High doses of vit. D. The initial dosage of vit. D is 50,000-300,000 units per day. Maintenance dosage is 20,000-150,000 units per day. This disorder differs from X-linked hypophosphatemic rickets by virtue of having an earlier onset with hypotonia, more severe hypocalcemia, less severe hypophosphatemia, and complete clinical and laboratory restitution on a relatively high dosage of vit. D [4].

3. Vit. D-Resistant Rickets (X- linked hypophosphatemic Rickets)

Abnormalities: mild to moderate growth deficiency (adult stature 130-160 cm).

Metabolic: Hypophosphatemia with diminished renal tubular reabsorption of phosphorus and calcium from gastro-intestinal tract; skeletal Rickets.

Dentition: Large pulp chamber with enamel hypoplasia; gingival and periapical infection; delayed eruption of dentition.

*Treatment***.** Vit. D taken in high dosage is potentially more harmful than the untreated disease. Combined treatment with 1-alpha hydroxyvitamin D and oral phosphate has been successful.

4. Albright Syndrome

5. Cleidocranial Dysostosis

6. Oculo-dento digital Syndrome

IV. Caries

Frequent in:

1. Cockayne Syndrome Abnormalities: growth deficiency with loss of adipose tissue by mid to late infancy; craniofacial (salt and pepper retinal pigmentation, optic atrophy). *Dentition: c*arious teeth.

*Performance***:** mental deficiency, weakness, moderate perceptive deafness [5].

Etiology: Autosomal recessive.

2. Gorlin Syndrome (Basal Cell Nevus Carcinoma Syndrome)

Abnormalities: craniofacial (bossing, broad nasal bridge); thorax; hands; skin; basal cell nevi over neck, upper arms, trunk and face**.**

Dentition: dyskeratotic cysts of mandible; occasionally maxilla; caries of teeth. *Performance:* variable mild to moderate mental deficiency.

Etiology: autosomal recessive.

3. Tricho-Rhino-Phalangeal Syndrome

Abnormalities: mild growth deficiency; facial; hair, nails; skeletal.

Dentition: small, carious teeth with dental malocclusion.

Etiology: autosomal dominant [6].

4. Oral-Facial-Digital Syndrome

*Abnormalities***:** oral-artial clefts in mid-upper lip, tongue, alveolar ridges, between premaxilla and lateral hard palate. Facial: hyplopasia of allar cartilages, lateral placement of inner canthi. Digital: clinodactily, syndactily, brachydactily.

Dentition: dental caries, anomalies of anterior teeth, absent lateral incisors. The ratio of female to male offspring of affected women has been 2:1.

V. Early loss of teeth

1. Oculo-Dento-Digital Syndrome. *Dentition:* enamel hypoplasia; early loss of teeth [7].

VI. Irregular placement of teeth.

Frequent in: Down Syndrome; Gorlin Syndrome; Hurler Syndrome; Morquio Syndrome; Pycnodyostosis; Tricho-Dento-Osseous Syndrome; Cohen Syndrome. *Dentition:* prominent maxillary central incisors; irregular placement of teeth.

Occasional in: Crouzon Syndrome; Ehler Danlos Syndrome; Maroteaux Lamy Syndrome.

VII. Late eruption of teeth (I , II , III, IV, V, VI, VII).

VIII. Neonatal teeth

Frequent in Chondroectodermal Dysplasia

IX. Other tooth anomalies.

Wide spaced in (Angelman Syndrome; Coffin Lowry Syndrome; Hay Wells Syndrome); Prominent teeth (Cohen Syndrome; Marshall Syndrome); Supernumerary (Gardner Syndrome; Oro-Facial-Digital Syndrome); Large teeth (XXX Syndrome).

Discussions

Our research analyzes a group of syndromes occurring in the bone and joint sys-

tem development anomalies, namely those that entail the dentition alteration, including facial bones development abnormalities [8].

This approach has been conducted at four essential levels:

1. **General,** morphologic and functional **aspects** (mental deficit degree included) for classification by categories of the bone development anomalies and determination of the kind of disease [1].

2. **The etiology** of such syndromes is either genetic (chromosomal, genetic), when lesions develop ever since the embryonic stage, or enzymopathic, case in which it entails storage diseases or absorption – reabsorption disorders, resulting in more or less severe bone modifications [9].

3. **The third approach criterion** and the one this work lies on, is the type, degree of severity and spreading of dental lesions [6].

Modifications of dentition have been classified according to the main type of disease: hypodontia, enamel hypoplasia, dental cavities, early loss of teeth, abnormal dental position, delayed dental eruption, neonatal teeth.

Although prevailing in certain syndromes, such modifications usually associate with a great diversity of dental disorders that may be found in diseases, such as: Cleidocranial dysplasia, Albright syndrome, Down syndrome, Osteogenesis Imperfecta etc.

In fact, such syndromes are most frequently found in the bone and joint system development anomalies, usually displaying dominant transmission (autosomal or X-linked).

4. And last, but not least, **the fourth** considered **criterion** focuses on the etiopathogenic and overall treatment that some of such diseases may take advantage from, as it supports the dental recovery effort, where necessary and appropriate [10].

Conclusions

Bone diseases of genetic origin also trigger modifications in dentition, as common alterations occur ever since the embryonic stage. Enzymopathies, which result in unmetbolized substances storage processes at the bone level, alter the facial bones and dentition as well. Determination of the type of bone disease, as well as the etiologic classification is absolutely necessary in order to anticipate dental problems and determine their prognosis and recurrence risk. Disorders in dentition development take on prevailing aspects in certain bone diseases, however in most cases such modifications coexist in the same syndrome. The same dental lesion may also be found in several congenital bone diseases. Etiopathogenic treatment of some of the above-described bone syndromes reduces the progress of bone lesions and supports the improvement effort and possibly the dental recovery.

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Correspondence to: Dr. Lenuta Malcea, 144 Mircea Blvd., Bl. MD7A, Sc.B, Apt.48, Constanta, Romania. E-mail: lmalcea@yahoo.com