

Oro-Facial Phenotypes Associated to Several Genetic Syndromes with Skeletal Dysplasia

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INTRODUCTION: In medical stomatological practice, genetic etiology accounts for an important part of dento-maxillary pathology. Hereditary involvement is a causative factor in isolated stomatological anomalies, but more often in complex syndromes. These syndromes are characterized by clinical heterogeneity, associating in phenotype oral manifestations and cranial, digital and scapulohumeral belt anomalies.

METHODS: The paper presents three clinical cases with specific phenotypes, but associating a pattern of anomalies of the cranio-facial structures derived from mesenchyme, dental agenesis, inclusions, anomalies of position and structure of the dental organ. The cases were diagnosed in the Medical Genetics Laboratory of the „Victor Babes” University of Medicine and Pharmacy Timisoara.

RESULTS: Case 1: Based on characteristic phenotypic manifestations: prominent nose, microphthalmia, broad first finger, with radial angulation, hyperextension, hypoplastic maxilla, first of the three cases was diagnosed with Rubinstein-Taybi syndrome. Cytogenetic analysis revealed a small deletion of the short arm of chromosome 16 (16p13).

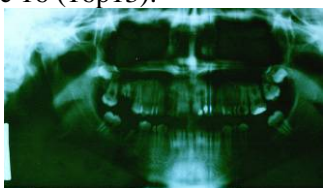


Fig. 1. Patient 1. Dento-maxillar anomalies: agenesis 3.8, 4.8, disrotation of 1.4.

The second case, a male patient, aged 15, was diagnosed with Floating-Harbor syndrome, with an autosomal dominant inheritance. Specific phenotypical manifestations of the syndrome found in this patient are: facial dysmorphism, hypoplastic mandibula, thin upper lip, dental agenesis, disrotations, macrodontia, clinodactyly of fifth finger, bipartite clavicle, the ossific nucleus of pisiform revealed delay of bone age of 3-3 ½ years. Cytogenetic analysis showed the deletion of the q24.1-q24.22 region on chromosome 8.



Fig. 2. Patient 2. Agenesis of 1.2 ; 2.2 ; 3.1 3.2; 4.2; 1.8; 2.8; 3.8; 4.8, dysrotation of 1.2 ; 4, rotation of 4.5, macrodontia of 1.1; 2.1; 2.3; 4.3, microdontia of 1.7.

The third case associates two different dysplasias: fronto-nasal and cleido-cranial dysplasia. The particularity of this patient's phenotype consists of association of facial aspects: hypertelorism, widow's peak, notched broad nasal tip—characteristic for fronto-nasal dysplasia, with skeletal and dental anomalies: hypoplastic maxilla, dysodontia, right clavicle hypoplasia with acromioclavicular disjunction, oblique right shoulder, short fifth finger – specific for cleido-cranial dysplasia. Dental anomalies found in this patient are: ectopic teeth, dysrotation, delayed eruption, crowded teeth.



Fig. 3. Patient 3. Orthopantomography - dental anomalies.

DISCUSSION & CONCLUSIONS: Compiling our data, the conclusion is that there is a strong correlation between the development of dental structures and oro-facial mesenchymal structures along with the development of skeleton. These pathological entities have direct consequences on the stomatognathic system, most of them being functional anomalies (mastication, deglutition and phonation disturbance, oral breathing), the disorders requiring preventive orthodontic approach and suitable management.

REFERENCES: ¹M. A. Smith., J.M Ferguson-Connor (2003) *Essential Medical Genetics*, Taylor&Francisc Inc.