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SHORT COMMUNICATION

P-17 IMPORTANCE OF THE ORTOPANTOMOGRAPHIC STUDY FOR THE DETECTION OF LIFE-THREATENING RARE DISEASES

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Keywords
"Rare diseases", "orthopantomography", "Marfan's syndrome", "Ehlers-Danlos's syndrome", "Gardner's syndrome", "Gorlin-Goltz's syndrome", "Total congenital hemihypertrophia", "Cowden's syndrome", "congenital dysqueratosis".

Introduction
A rapid diagnostic of some Rare Diseases (RD) can save the patent's life. It is well known that some life-threatening Rare Diseases present oral and craniofacial malformations.
If the RD patient presents obvious malformations, either craniofacial or systemic, the diagnosis can be made at birth or in the first months/years of life. However, some syndromic entities can remain undetected until the oral clinical signs are observed by the dentist. The awareness of the dentist about such phenotypical traits is thus essential. Orthopantomography –a simple and cheap diagnostic image- can be the instrument for revealing a previously undetected RD.
The aim of our study is to review the oral and craniofacial phenotypical traits of the gravest and life-threatening RDs to establish its orthopantomographic evidences.

Material and Methods
A Medline search was conducted including the following search-term combinations: "orthopantomography" or "oral signs/symptoms" and "Rare diseases", "Marfan's syndrome", "Ehlers-Danlos's syndrome", "Gardner's syndrome", "Gorlin-Goltz's syndrome", "Loeys-Dietz's síndrome", "Total congenital hemihypertrophia", "Cowden's syndrome" and "congenital dysqueratosis". The terms related to the syndrome's names were chosen following the ORPHANET nomenclature. Given the limited number of cases published, no additional set of criteria was defined to subsequently filter the resulting articles.

Discussion
The Gorlin-Goltz's syndrome can be diagnosed radiologically because of the presence of maxilar keratokysts. The keratokysts constitute a life-threatening pathology because they can host malignant tumors such as intrakystic ameloblastomas or perikystic fibrosarcomas. Moreover this clinical entity is related to the presence of meduloblastomas or multicentric skin basal cell carcinomas (1)
Gardner's síndrome can be diagnosed radiologically because of the presence of osteomas in skull, face and jaws, as well as odontomas, folicular kysts, supernumerary and/or included teeth. This syndrome is associated to the familial adenomatous polyposys, with a 100% risk of malignant transformation (2,3,4).
Total congenital hemihypertrophia is evident because of the hypertrophy of both the calcified and the non-calcified tissues of half the oral cavity, the face and the rest of the body. In the affected oral area, the dental remplacement is accelerated, and dental agenesis and microdontias can be found. This entity is associated to malignant pathology, such as nefroblastomas and hepatoblastomas (2, 3,4)
Entities that affect the fibrous componentes of connective tissues, such as Marfan's syndrome and Ehlers-Danlos's syndrome are related to a life-threatening pathology such as the acute aorctic dissection in young patients

(5,6,7,8,9). The common embryologic origin in the neural crest of the craniofacial bone structures and the thoracic big vessels can explain the simultaneous presence in their patients of craniofacial and vascular clinical signs (10). Plurisystemic syndromes include oral clinical manifestations that can be verified by an orthopantomography. The oral phenotype of the Marfan's syndrome includes supernumerary teeth, maloclusive signs, pulpolythes, enamel hypoplasia, deformation of the teeth's roots or pulpar alterations. Ehlers-Danlos's syndrome oral aspects are also supernumerary teeth or hypodontia, added to haemorraghic bullae, parodontitis, caries, and TMJ hypermobility. (5,6,7,8,9).
Cowden's syndrome, Loeys-Dietz's syndrome and congenital dysqueratosis are also life-threatening pathologies presenting oral phenotypical traits, but related only to smooth tissues. These signs and symptoms are thus better explored by a clinical expert than in an orthopantomography (10,11,12,13,14).

Conclusions:
Numerous life-threatening rare diseases present oral phenotypical traits that can be detected by an orthopantomography. The dentist's awareness about the oral signs and symptoms of rare diseases, "a fortiori" the life-threatening ones, can contribute to their precocious diagnosis. In order to contribute to the rapid diagnostic of rare diseases, dentists must be aware of their craniofacial phenotypical traits.

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