

Oral pathology in a group of Mexican patients with genetic diseases

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ABSTRACT

Without considering infectious and traumatic diseases, the great majority of oral cavity diseases have a genetic base, in some cases identifiable, in others not. For the stomatologists it is of great importance to know the clinical characteristics and type of alteration that go with genetic etiology syndromes to be able to offer patients an adequate multidisciplinary treatment. Objective: Intentional search and description of oral pathology in patients with diverse genetic diseases.

Study design: An observational and descriptive 4 month study of 62 patients from the Genetics Department of the National Institute of Rehabilitation, was done. Taken into consideration, aside from oral manifestations and genetic disease, were age, sex, consanguinity and inbreeding. The majority of patients who have genetic pathology do not have curative treatment, but they can receive other treatments to improve their quality of life, among these are dental treatments.

Results: The more common diseases we found were hereditary peripheral neuropathies, skeletal dysplasias, limb malformation and muscular dystrophies. Diverse features were described, registered and grouped according to their location.

Conclusions: Presently it is important to look for the genetic etiology of all diseases to seek specific treatments and prevent them. This will change the practice of medicine and dentistry.

Key words: *Oral pathology, genetic pathology, dentistry.*

RESUMEN

Dejando de lado la patología infecciosa y traumática, la gran mayoría de las patologías odontológicas, tienen una base genética, en algunas de ellas identificada, en otras no. Para los estomatólogos es de gran importancia conocer las características clínicas y el tipo de alteración que acompañan a los síndromes de etiología genética, para poder ofrecer a los pacientes un tratamiento apropiado y multidisciplinario.

Objetivo: Búsqueda intencional y descripción de la patología bucal en pacientes con diversas enfermedades genéticas.

Diseño del estudio: Se realizó un estudio observacional y descriptivo reuniendo a 62 pacientes de la consulta de Genética del Instituto Nacional de Rehabilitación durante 4 meses. Se tomaron en cuenta, además de las manifestaciones bucales y la enfermedad genética relacionada a las mismas, el sexo, la edad, la presencia o no de consanguinidad, y endogamia, así como la localización de la manifestación bucal. La mayoría de los pacientes que presentan patología genética no tienen tratamiento curativo, pero sí podrán realizarse otros tratamientos para mejorar su calidad de vida, entre estos se cuentan los diversos tratamientos odontológicos.

Resultados: Los padecimientos diagnosticados con mayor frecuencia fueron neuropatías periféricas hereditarias, displasias esqueléticas, malformaciones de miembros y distrofias musculares. Se describieron diversas manifestaciones que fueron registradas y agrupadas de acuerdo a su localización.

Conclusiones: En la actualidad se busca conocer la etiología genética de todos los padecimientos para buscar tratamientos específicos y prevenir su aparición, hechos que revolucionarán la práctica médica y la odontológica.

Palabras clave: Estomatología, patología bucal, patología genética.

INTRODUCTION

Without considering the infection and traumatic diseases, the great majority of the oral diseases have a genetic base, in some of them we know it but in others we do not; but for their characteristics, signs and symptoms we can assume they are diseases with a genetic origin (1-3).

For the dentist it is very important to know the clinical characteristics and the possible alterations that are part of the genetic syndromes, to offer patients multidisciplinary and the best possible treatments. Many of these syndromes can be diagnosed by their oral characteristics, which state the importance of the dentist having full knowledge of them (4).

As infectious diseases are controlled, the percentage of genetic illnesses is greater, as can be seen in third level pediatric hospitals in Mexico and the rest of the world. Many of the genetic pathologies that go with malformations, present alterations in the maxilofacial region (5).

Knowledge of genetically based diseases is growing at a very fast pace. Although it seems there is a promising future in this area, genetic integration in every day medical practice still presents difficulties.

The object of this study is the intentional search and description of oral pathology in patients with diverse genetic diseases.

PATIENTS AND METHODS

An observational and descriptive study of all the patients from the genetic department of the National Institute of Rehabilitation (INR) in Mexico City, during 4 months; from 15 August to 15 December 2005, was done.

There were compiled data of genetic disease, oral characteristics, age, sex, consanguinity, inbreeding and the place of the oral findings.

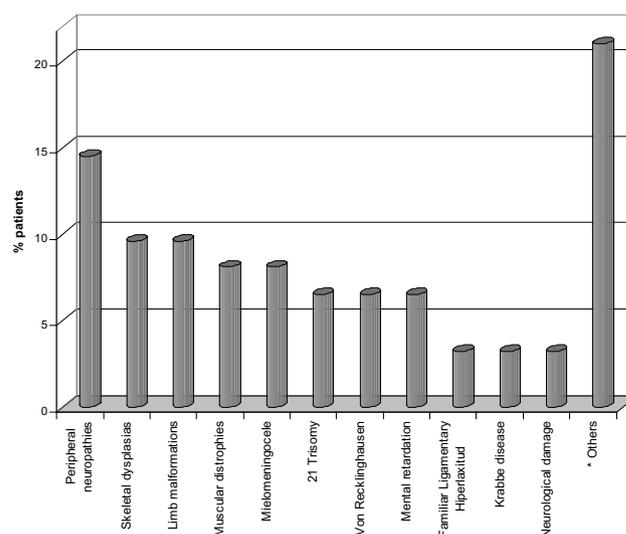
For each of these patients, a genetic and oral clinical history, clinical photos, radiological studies and other kind of studies were made depending on the probable or definite diagnosis. This was done either to confirm diagnoses or/and to look for the alterations normally present in these illnesses. The oral revision and the description of the features were carried out by both, the dentist and the oral pathologist, reason why we did not make the kappa index, we did only descriptive statistics.

RESULTS

62 patients were examined, 36 were male (58.1%) and 26 were female (49.1%). The patients age was between 1 month and 43 years, the average was 8.4 years, mode and median 5 years.

Only 1 patient referred consanguinity (1.61 %). Inbreeding was referred in 2 cases (3.22%) and in 9 patients it was con-

sidered probable; the other 51 patients (82.35%) denied it. The most frequent diagnoses were peripheral hereditary neuropathy (9 patients) (6), skeletal dysplasias (6 patients), limb malformation (6 patients), muscular dystrophies (5 patients) (7), myelomeningocele (5 patients), 21 trisomy (4 patients), Von Recklinghausen disease (NF1) (4 patients) (8), mental retardation (4 patients). Other disorders found were ring constriction syndrome, incontinentia pigmenti, Cockayne syndrome, ligamentary hyperlaxitud, among others (9-10). (Fig.1)



Ring constrictions, Klinefelter, Artrogriposis multiplex congenita, Möebius, Hipomelanosis of Ito, Silver Russell, Incontinentia Pigmenti, Cornelia de Lange, Cockayne, collagenopathy, acrorenal, Sotos and pterigium multiplex syndromes.

Fig. 1. Frequency of the diseases founded.

In the oral exploration, different features were observed that were registered according to the place they were located, dental occlusion was also evaluated. There were some patients without oral features and some others with more than one. We found 4 patients with features in the oral mucose, 7 in palate, 7 in tongue, 40 in teeth, 17 in the gingiva, 34 in the occlusion and 6 with diverse features. 18 out of the 62 patients had no oral manifestations. (Tabla 1)

Table 1. Regions with oral features.

Regions	Features
Oral mucose: (4)	Fordyce granules 4/4.
Palate: (7)	Spots of neurofibromatosis 1/7 High palate 4/7 Narrow palate 4/7 Scar of cleft palate 1/7 Palate fistula 1/7.
Tongue: (7)	Bifid tongue 1/7 Geographic tongue 2/7 Fissured tongue 3/7 Ankyloglossia 2/7 Macroglossia 1/7.
Teeth: (40)	Focal or localized macrodontia 2/40 Focal or localized microdontia 1/40 Fusion/Gemination 2/40 Delayed eruption 1/40 Hypodontia 4/40 Enamel hypoplastic 2/40 Short roots 1/40 Caries 36/40
Gingiva: (17)	Gingivitis 16/17 Periodontal disease 1/17
Occlusion: (34)	Crowding lower dental 9/34 Crowding upper dental 5/34 Dental malposition 17/34 Anterior crossbite 2/34 Unilateral posterior crossbite 2/34 Anterior open bite 3/34 Border to border bite 2/34 Straight terminal plane 5/34 Mesial exaggerated terminal plane 3/34 Class II of Angle 2/34 Class III of Angle 1/34
Others: (6)	Mandibular hypoplasia 2/6 Maxillary hypoplasia 1/6 Hipertrophic tonsils 1/6 Comissural lip pits 1/6 Appendix frenum 1/6.
Without features: (18)	18/62
Note: Due to the fact that some patients presented more than one feature the total number is larger than the number of patients.	

DISCUSSION AND CONCLUSIONS

The present work allowed us to establish the importance of the complete and interdisciplinary study of these patients. Moreover, this is more relevant when we study patients with a genetic disease, because the morphogenesis or the structural proteins and their functions depend on genetic factors, which act in multiple levels of the human body (12, 13). Taking into consideration that there are usually many

affected tissues, some times for a single gene, we can see the importance of looking for features in the different body areas (12, 14). The mouth is made up of different tissues, which is the reason it is very frequent to find oral pathologies in patients with genetic diseases. Some oral structures are derived from the ectodermic layer, which is why it is very important, in the patients with genetic disease with neuroectodermic features, to carefully check patients looking for possible oral pathological characteristics (14,15).

The patients we studied have diseases that need rehabilitation therapies, specially in the muscular and skeletal areas; that is why the majority of them have the same oral features and in the same proportion of the general population (2,16). However, although in less quantity, we already have patients with pathological oral features such as mandibular hypoplasia, hypodontia, short roots, conic teeth, in patients with Hipomelanosis of Ito, Sotos syndrome, Cockayne and Incontinentia pigmenti (IP2) syndromes (10,11).

The importance of studying the patient in an integral way is to know what we can offer him depending on the anomalies detected. Many patients with genetic pathology can not be cured, but we can give them different treatments to improve their quality of life, such as orthodontic, orthopedic, surgeries, restoration and preventive treatments, etc (16,17).

For all the above, it is of special importance, that the dentist know the basic genetic principles and be able to detect these features in his patients as well as the risk of developing comorbid pathology for being "special" patients, as periodontal disease and caries, that are more frequent in some patients because of the difficult to carry out the oral hygiene and in others because these features could be part of the genetic pathology.

The therapeutic possibilities for hereditary diseases include pharmacological therapy, dietary modification, metabolic manipulation, environment modification, gene therapy, reconstructive surgery, physical, occupational and language therapies (18).

Presently it is important to look for the genetic etiology of all diseases to have specific treatments and prevent them, conditions that will change the practice of medicine and dentistry (19).

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