

Orthopedic Clinical Manifestations of Ectodermal Dysplasia. Case Presentation

Espinosa LCB¹, Estupinan LMM², Aparicio LM³, Gonzalez JCC⁴, Pazos CP^{5*} and Plain LD⁶

¹I Degree Specialist in Orthopedics and Traumatology, Assistant Professor, Provincial General University Hospital “Mártires del 9 de Abril”, Sagua la Grande, Villa Clara, Cuba

²Doctor in Medical Sciences, II Degree Specialist in Orthopedics and Traumatology, Full Professor, Provincial General University Hospital “Mártires del 9 de Abril” Sagua la Grande, Cuba

³3rd year resident of Orthopedics and Traumatology, Provincial General University Hospital “Mártires del 9 de Abril” Sagua la Grande, Cuba

⁴IIDegreeSpecialist in Orthopedics and Traumatology Provincial General University Hospital “Mártires del 9 de Abril” Sagua la Grande, Cuba

⁵Specialist of I and II Degree in Comprehensive General Medicine, Faculty of Medical Sciences of Sagua la Grande, Cuba

⁶Resident of 3rd year of Orthopedics and Traumatology, Provincial General University Hospital “Mártires del 9 de Abril”, Sagua la Grande, Villa Clara, Cuba

*Corresponding author:

Claribel Plain Pazos,
Faculty of Medical Sciences of Sagua la Grande,
Villa Clara, Cuba,
E-mail: claribelpp@infomed.sld.cu

Received: 15 Apr 2021

Accepted: 10 May 2021

Published: 15 May 2021

Copyright:

©2021 Pazos CP. This is an open access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and build upon your work non-commercially.

Citation:

Pazos CP. Orthopedic Clinical Manifestations of Ectodermal Dysplasia. Case Presentation. Ann Clin Med Case Rep. 2021; V6(14): 1-4

Keywords:

Ectodermal dysplasia; Orthopedic clinical manifestations

1. Resume

Ectodermal dysplasias represent a heterogeneous group of alterations, characterized by the abnormal development of embryological derivatives of the ectoderm. We present a patient who, upon examination of the osteomyoarticular system, presents intense rotation of the lower limbs with the toes backwards, which the patient performed without difficulty, in an unforced, voluntary and painless way. Orthopedic clinical manifestations are infrequent in skeletal dysplasias, lower limb disorders even rarer, which is why we present these striking alterations, a fundamental objective of the study.

2. Introduction

Ectodermal dysplasias represent a heterogeneous group of alterations, characterized by the abnormal development of embryological derivatives of the ectoderm. More than 150 types have been described, all of them hereditary, with genetic heterogeneity and great variability in their expression, mostly linked to the X chromosome or autosomal recessive. In a lesser frequency, the autosomal

dominant one, the hypohydrotic form is the most common [1]. It is estimated that 7 out of every 100,000 newborns, without predominance of race, present some form of ectodermal dysplasia [2], and the proportion between genders is five men for one woman [3].

In general, they are differentiated into 2 groups of disorders: one characterized by aplasia or hypoplasia of the ectodermal derivatives, which fail in their development and differentiation due to the absence of specific reciprocal signals between ectoderm and mesenchyme. The other, the most striking feature is palmoplantar keratoderma, which occurs in association with other manifestations when other highly specialized epithelia are affected [2].

The diagnostic triad is characterized by nail dystrophy (onychodysplasia), alopecia or hypotrichosis (sparse, thin, and light hair on the scalp and eyebrows), and palmar-plantar hyperkeratosis that is often accompanied by a lack of sweat glands (hypohidrosis) and the partial or complete absence of deciduous and / or permanent teeth [3].

Orthopedic clinical manifestations are infrequent in skeletal dys-

plasias, lower limb disorders even rarer, which is why we present these striking alterations, a fundamental objective of the study.

3. Case presentation

An 18-year-old male patient with a history of having suffered from Server's disease, for which he was treated in the health area, resolving the condition without sequelae. He presented dental alterations due to the absence of incisors in the lower arch, being assisted in specialized stomatological services where dental prostheses were implanted. He also refers to heat intolerance. He denies the occurrence of repetitive fractures during childhood or delayed learning, he practiced contact sports such as karate, without major physical problems.

His family history is an older brother than him, a carrier of ectodermal dysplasia diagnosed by genetic consultation, who presented dental alterations due to delayed teeth and skin with areas that did not present sweating. Father, mother and the rest of his relatives with an apparent health history.

When he was evaluated during a routine check-up of the general military service, a physical examination was carried out and the following alterations were found:

Skin and mucous membranes: smooth areas on the thighs and trunk with little sweating.

Faneras: incomplete eyebrows and some areas of the face without beard hairs.

Oral examination: Absence of incisors in the lower arch.

Examination of the osteomyoarticular system: patient in a standing position performed external hyper-rotation of the lower limbs with the toes back, without difficulty, in a non-forced, voluntary and painless way (Figure 1), not hypermobility of the joints. Normal gait and squat. Light cavus foot, without plantar calluses. Fabere Patrick and Bado maneuvers of both normal lower limbs. Forced movements in normal internal and external rotation. When examining the anterior part of the coxofemoral joints, the heads of the femur were not palpated, considering the possibility of subluxation of the same; In supine decubitus, it was not possible to obtain the aforementioned backward rotation position of the lower limbs, that is, this hyper-rotation is only possible while standing. This resulted in the impossibility of performing computerized axial tomography in our case, only the serial radiographs that appear in this work (Figure 2) and correlated with the photographs obtained.



Figure 1: Características clínicas ortopédicas en el miembro inferior del paciente. Nótese: característica de los miembros inferiores en posición normal (1), se observa como puede rotar los dos miembros hacia detrás, más de 90° (2), observe como rota totalmente el miembro derecho (3) y el miembro izquierdo (4).



Figure 2: Características radiológicas ortopédicas en la pelvis y las caderas. Nótese: radiografía de pelvis ósea con caderas en posición normal (1), se observa al rayos X como puede rotar los dos miembros hacia detrás, más de 90° y la imagen del cuello femoral se superpone (2), observe como rota totalmente el miembro derecho (3) y el miembro izquierdo (4).

4. Discussion

The brother of the patient presented is a carrier of ectodermal dysplasia, he was diagnosed by genetic consultation, due to the presence of few characteristic elements of the disease (delay in the emergence of teeth and skin with areas that did not present sweating) the above, led to suppose the authors in an autosomal dominant inheritance with incomplete penetration phenomenon [3].

To study the case, the patient was referred to Stomatology, Dermatology and Otorhinolaryngology consultations. No alterations of the throat, nose or ears were found, a common issue to that raised by other authors [4].

One of the main characteristics of Skeletal Dysplasias is dental alterations [5], which are very diverse and were present in this case. Mutations of the morpho-genetic regulatory genes involved in determining the position and early development of dental organs (homeobox genes), alter dental morpho-differentiation, have pleiotropic effects in different organs, hence the affectations present in different tissues [6]. It was noted that the patient had only the absence of the lower incisors with the rest of the normal dental arch, which did not coincide with other authors, who found more dental involvement in the patients studied [7,8].

Given by the alterations of the skin, sweat glands and dentition, the

authors consider hypohidrotic ectodermal dysplasia as a diagnosis [3].

The clinical diagnosis is always accompanied by imaging studies and, depending on the age at which it is diagnosed, the treatment will always be aimed at reestablishing and improving the quality of life.

In several large studies carried out, authors provide multiple clinical manifestations, but there are few that address orthopedic manifestations [3,9,10], especially articular ones, which affect the quality of life of patients.

Despite the fact that in Cuba a multidisciplinary consultation is established for the diagnosis and follow-up of these patients where skin surveillance is carried out by dermatologists and orthodontic problems by the team's stomatologists; a social worker, a psychologist and a clinical geneticist also participate, there is no presentation of the orthopedic clinical conditions of these pathologies.

5. Conclusions

Ectodermal dysplasia occupies a relevant place in medical-dental and imaging practice; On the basis of the present investigation, it is of great importance to offer a professional and multidisciplinary approach that allows knowing its etiology, clinical presentations; in this way, accurate and accurate diagnoses can be reached, using

reliable and safe imaging means, as well as treatments that improve the quality of life.

References:

1. Ramírez M, Jaimes L, Pieruccini JF, et al. Displasia ectodérmica: un reporte decaso. *RevEstomatolHer*. 2016 [citado 14/06/2018]; 3(26): (Aprox.3p.).
2. Luna Ceballos EJ, Domínguez Pérez ME, Sainz Padrón L, Rodríguez Acosta Y. Displasia ectodérmica hipohidrótica autosómica dominante asociada ahipoparatiroidismo. *RevMéd Electrón*. 2019 [citado:24/2/2021]; 41(4): (Aprox.8p.).
3. Schautz CG et al. Ectodermal Dysplasia in child: multidisciplinary management. *BrazDentSci*. 2019 Apr/Jun [citado:24/2/2021]; 22(2): 289-95.
4. Vera Sáez Benito MC, Álvarez Ballano D, De Arriba Muñoz A. Hipoparatiroidismo y trastorno del desarrollo sexual: una asociación inusual. Carta al editor. *Med Clínica*. 2017 [citado 14/06/2018]; 150(1): (Aprox.6p.).
5. Sfeir E, Nahass MG, Mourad A. Evaluation of Masticatory Stimulation Effect on the Maxillary Transversal Growth in Ectodermal Dysplasia Children. *Int J ClinPediaterDent*. 2017[citado 10 mar 2018]; 10(1): 55-61.
6. Dean JA, Turner EG. Eruption of the teeth: local, systemic, and congenital factors that influence the process. In: Dean JA, editors. *McDonald and Avery's Dentistry for the Child and Adolescent*. 10th ed. St. Louis, MO: Elsevier Mosby; 2016.
7. Velazque Rojas L, Dalben da Silva G. Displasia ectodérmica hipohidrótica. Características clínicas y radiográficas. *RevOdontMex [Internet]*. 2015 [citado 24/02/2021]; 19(4): (Aprox.8p.).
8. Bassi JC, Antunes Santos AM, Kerber Tedesco T, et al. Rehabilitación oral enpacientes con displasia ectodérmica: un año de seguimiento. *RevDig Odontología*. 2016 [citado 24/02/2021]; 18(2): (Aprox.6p.).
9. Rodríguez MM. Etiología, manifestaciones clínicas y estudio por imágenes de la displasia ectodérmica: una revisión de la literatura. *RevCientOdontol (Lima)*. 2018; 6(2):236-47.
10. Santana Hernández EE. Comportamiento de las displasias ectodérmicas en la provincia de Holguín. Cuidados de la piel y protección social. *Correo Científico Médico (CCM)*. 2019; 23(4).