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Orthopedic Clinical Manifestations of Ectodermal Dysplasia. Case Presentation

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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 (onchodysplasia), 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.

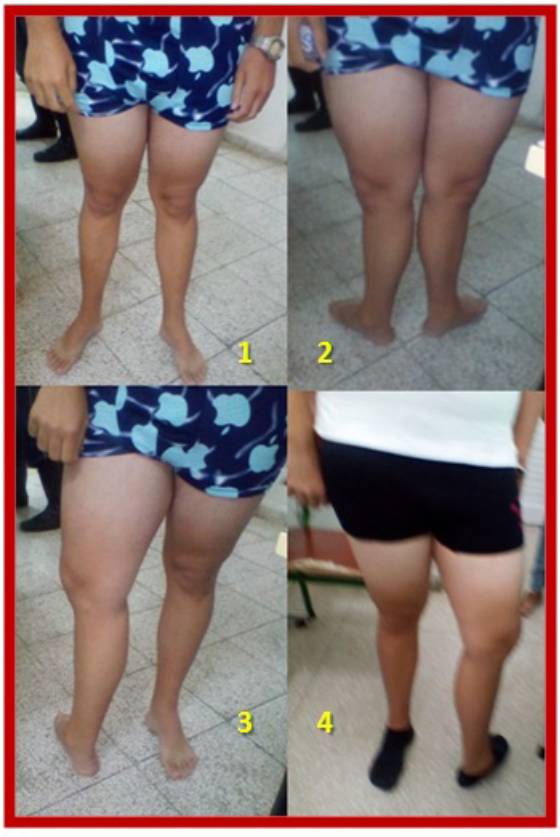

Figure 1: Características clínicas ortopédicas en el miembro inferior del paciente. Nótese: característica de los miembros inferiores en posición normal (1), se observa como puede rotar los dos miembros hacia detrás, más de 90º (2), observe como rota totalmente el miembro derecho (3) y el miembro izquierdo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

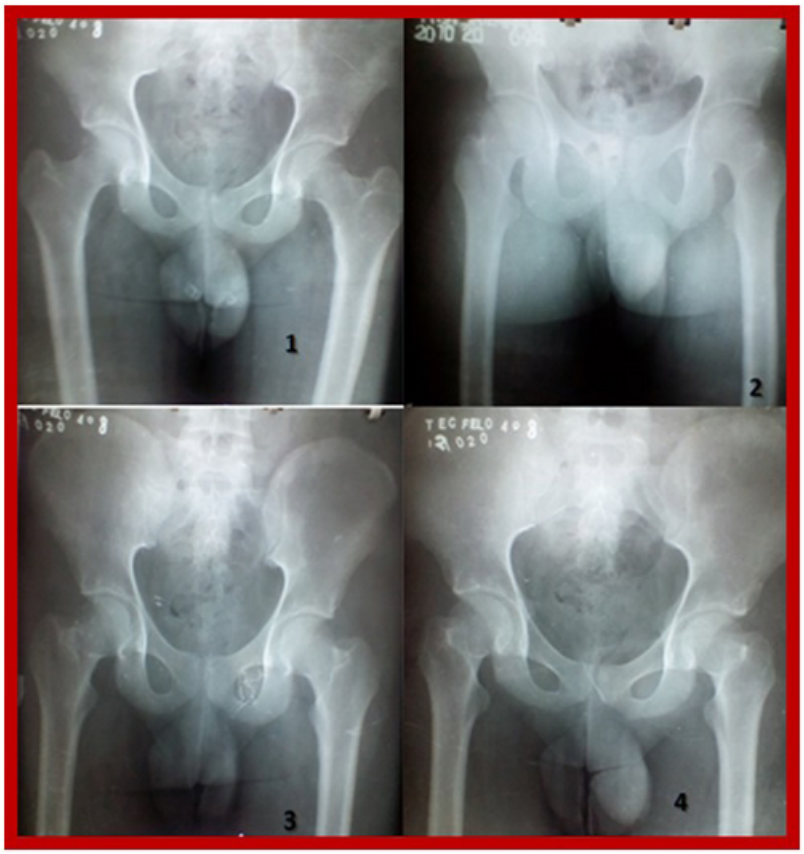

Figure 2: Características radiológicas ortopédicas en la pelvis y las caderas. Nótese: radiografía de pelvis ósea con caderas en posición normal (1), se observa al rayos X como puede rotar los dos miembros hacia detrás, más de 90º y la imagen del cuello femoral se superpone (2), observe como rota totalmente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

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