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Síndrome de Escobar: A propósito de un caso

Escobar Syndrome About a case

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Palabras claves:
Autosómico, pterigium, contractura, cariotipo, displasia.

Resumen

Introducción: El Síndrome de Escobar o pterigium múltiple no letal, es una entidad poco frecuente con un patrón de herencia autosómica recesiva. Su etiología es desconocida, pero clínicamente se caracteriza por presentar pterigium múltiple, contracturas musculares y alteraciones esqueléticas. Las primeras descripciones, se hicieron a principios del siglo XX por Bussiere y Frawley, pero fue Víctor Escobar en 1978 quien lo estudió y lo describió de una forma más amplia. **Objetivos:** Describir características clínicas y diagnósticas del síndrome de escobar. **Metodología:** En el presente trabajo, se describe un caso, que cuenta clínicamente con alteraciones osteomusculares que limitan parcialmente su calidad d e vida. No presenta complicaciones pulmonares. Actualmente se encuentra en investigación el cariotipo y cirugías correctivas de caderas y pies. **Resultados:** En el presente caso clínico, sus principales alteraciones son la escoliosis, pterigiones múltiples, camptodactilia, displasia de cadera y el pie equino-valgo bilateral, lo cual limita parcialmente su calidad de vida. La paciente no presenta restricción pulmonar, a pesar de ser la complicación más frecuente, indicador de buen pronóstico. Nuestro caso se encuentra en investigación del cariotipo y cirugías correctivas en cadera y pies. **Conclusiones:** Es una enfermedad genética con mecanismo de herencia autosómica recesiva cuya fisiopatología aún se desconoce. El diagnóstico se confirma por medio de pruebas genéticas que evidencien la mutación del den CHRNG, sin embargo, su ausencia no descarta el Síndrome de Escobar. **Área de estudio general:** Medicina. **Área de estudio específica:** Pediatría. **Tipo de artículo:** Caso clínico.

Keywords:
Autosomal, pterygium, contracture, karyotype, dysplasia.

Abstract

Introduction: Escobar syndrome or non-lethal multiple pterygiums is a rare entity with an autosomal recessive inheritance pattern. Its etiology is unknown, but clinically it is characterized by multiple pterygiums, muscular contractures, and skeletal alterations. The earliest descriptions were made early in the 20th century by Bussiere and Frawley, but it was Victor Escobar in 1978 who studied it and described it more broadly. Objectives: Describe clinical and diagnostic

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characteristics of Escobar syndrome.**Methodology:** In the present study, a case is described, which has clinical features with musculoskeletal alterations that partially limit its quality of life not pulmonary and currently complications. She engages in karyotype research and corrective hip and foot surgeries. Results: In the present clinical case, its main alterations are scoliosis, multiple pterygiums, camptodactyly, hip dysplasia and bilateral equine-valgus foot, which partially limits their quality of life. The patient does not present pulmonary restriction, despite being the most frequent complication, an indicator of a good prognosis. Our case is undergoing karyotype investigation and corrective hip and foot surgeries. Conclusions: It is a genetic disease with autosomal recessive inheritance mechanism whose pathophysiology is still unknown. The diagnosis is confirmed by means of genetic tests that show the mutation of the CHRNG den, however, its absence does not rule out Escobar Syndrome. General study area: Medicine. Specific area of study: Pediatrics. Article type: Clinical case.

Introduction

Escobar syndrome or non-lethal multiple pterygium syndrome is a rare condition. Generally, the inheritance of this pattern is usually autosomal recessive, although a case of autosomal dominant inheritance has also been documented (1). It is distinguished by the presence of multiple pterygium in the neck and joints, joint contractures, intrauterine and postnatal growth restriction, accompanied by skeletal alterations and other malformations that may vary in frequency. Other manifestations include decreased movement, facial weakness, respiratory difficulty, vertebral abnormalities, scoliosis, finger abnormalities and cryptorchidism, among others. The disease is a non-lethal variant of multiple pterygium syndrome (2).

Non-lethal multiple pterygium syndrome or Escobar-type variety is characterized by multiple pterygium and congenital contractures. Few autosomal dominant cases have been reported and it also shows variable expressivity among those affected. It is associated with mutations in the CHRNG gene located at locus 2q37. Pterygia can be located in the neck, axilla, antecubital, popliteal and intercrural regions and become more evident over time, causing the appearance of joint contractures. In addition to congenital

contractures, various malformations and dysmorphisms such as ptosis, micrognathia, kyphoscoliosis, syndactyly, camptodactyly and short stature, among others, are present (Table I). Intellectual capacity is not affected. In many cases, these patients present complications due to various respiratory problems, possibly secondary to kyphoscoliosis and small thorax, which causes significant morbidity. As a result, approximately 6% of patients die in the first year of life (2).

Multiple pterygium syndrome, initially identified by Bussiere in 1902 (3), was characterized by Frawley in 1925 (4). In 1976, Gorlin et al. classified it as an autosomal recessive entity. In 1978, Escobar et al. carried out a comprehensive review and a detailed description of the syndrome, which led to the designation of a less severe variant as Escobar variant or Escobar syndrome (5).

It is a very rare entity, there are 100 reported cases around the world,This theory was initially proposed in 1999 (6).

Currently in Ecuador, there is no published case report of Escobar Syndrome, which is why we were driven and motivated to carry out this work. It is important to emphasize that, as it is a rare pathology worldwide, its diagnoses are complex. By sharing this research we will be able to reach a larger population and therefore, be a scientific and statistical contribution at the level of Latin America and the world.

Methodology

We present a documentary, analytical and correlational investigation of a patient diagnosed with Escobar Syndrome at the General Hospital of Portoviejo (IESS). A review of the clinical case was carried out in the medical history of the aforementioned hospital, including the clinical and imaging characteristics important to establish the definitive diagnosis and differential with other entities.

In this paper, we describe a case of a patient with clinical musculoskeletal disorders that partially limit his quality of life. He does not have any pulmonary complications. Karyotype and corrective surgeries of hips and feet are currently being investigated.

Etiology

The etiology of this condition is unknown, however, it is associated with homozygous or compound heterozygous mutations in the CHRNG gene (6). This gene is responsible for encoding the gamma subunit of the acetylcholine receptor (AChR) (7), which is present in the fetus until approximately week 33 of gestation.

Clinic

In the prenatal stage it is difficult to find data suggesting that the fetus is affected by the

non-lethal form of the disease. The clinical expression is very variable and, in the most severe form, which is lethal multiple pterygium syndrome, uterine growth retardation may occur (8) and about half of the cases may present hydrops or cystic hygroma (9), among other conditions.

Clinically, it presents multiple pterygium, which are defined as skin folds that pass through one or more joints limiting their range of motion. These can be located in the neck, armpit, antecubital, popliteal and intercrural regions and become more evident over time, causing the appearance of joint contractures. In addition, this syndrome is associated with two other structural abnormalities: a vertical talus and congenital lordescoliosis. (10)

In addition to congenital contractures, there are various malformations, expressionless appearance or lack of emotion and dysmorphia (11), which are divided into:

Minor malformations

- Low size
- Low auricular implantation
- Expressionless face
- Hypertelorism
- Micrognathia
- Camptodactyly
- Syndactyly (12, 13)

Major malformations

- Scoliosis
- Equine foot
- Pterygium multiple
- Heart murmur
- Cleft lip and palate
- Cryptorchidism (14)

It may be accompanied on some occasions by hip dislocation, absence of patellas, inguinal hernias, platyspondyly, and spina bifida occulta.

Intellectual capacity is normal. Complications that these patients may present include various respiratory problems, possibly secondary to kyphoscoliosis and a small thorax, which causes significant morbidity, since around 6% of patients die in the first year of life.

Diagnosis

It is performed clinically at birth and in other cases it can be diagnosed by uterine ultrasound and then confirmed in the neonatal period (14).

Prenatal diagnosis of this pathology is difficult due to the limited visualization of the pterygium; however, the presence of cystic hygroma, hydrops, and contractures in the extremities in marked flexion and without movement guide the diagnosis.

This syndrome is therefore confirmed by a positive sequence analysis for the CHRNG gene. However, there are individuals who present clinical data characteristic of this syndrome in whom mutations in this gene have not been identified, which suggests that there may be locus heterogeneity.

Differential Diagnosis

- Arthrogryposis Multiplex Congenita
- Pterygium popliteus
- Pterygium Isolated
- Noonan syndrome
- Turner Syndrome (12)

Clinical Examination

After the clinical history has been taken, with important details of the family history, prenatal and neonatal history, a thorough, correct and detailed physical examination is carried out, mainly of the head, neck, thorax, abdomen, cardiac system and musculoskeletal system. In addition, the respective assessment of anthropometric measurements is carried out, with a description of the patient's position, limbs and joints that are involved and the amount of connective tissue and muscle mass observed.

During the physical examination of the patient, the physician should carefully document the exact position and degree of movement of each contracture and whether any other abnormalities are observed.

Complementary Exams

Although not all patients with Escobar Syndrome have mutations in the CHRNG gene, it is advisable to perform the molecular study, especially in cases where an autosomal recessive inheritance pattern is suspected.

Identification of the mutation will allow for better genetic counseling for family members and opens up the possibility of performing a prenatal diagnosis in subsequent pregnancies.

X-rays play an important role in this pathology, both in the diagnosis, as well as in the evolution, prognosis and treatment. They assess the presence of hip dislocations, patella dislocations and scoliosis.

Muscle biopsy guides the diagnosis and the electrophysiological study allows to assess muscle activity and determine the specific location of the affectation.

When there is suspicion of neurological alteration, an Electroencephalogram or Magnetic Nuclear Resonance is indicated.

Evolution

Patients affected by this syndrome receive multidisciplinary management for their conditions. Some of them have difficulties swallowing and even a difficult airway to manage in intubation procedures (15).

The joints of the shoulders, elbows and hands must to be attended tometiculously with the aim of promoting correct function and mobilization of their joints, through physical therapy and occupational therapy.

It is difficult to give a prognosis during the first years of life, however, at 3 to 4 years of age it is easier, since, with physiotherapy, orthoses and surgeries, most cases manage to develop well.

Treatment

Patients with Escobar Syndrome should receive timely and multidisciplinary management aimed at the malformations they present. Treatment is based primarily on physical therapy, occupational therapy and surgery.

- In hip contractures less than 30°, conservative treatment with physiotherapy.
- In knee contractures of 25° or less without limitation in walking, treatment is conservative with lengthening exercises.
- Contractures of 25° to 90°, treatment options include physical therapy, “z” plasty, excision of the fibrous band and even lengthening of all structures to the extent that the neurovascular package allows, and then applying serial casts for 6 to 8 weeks.
- Contractures of 90° or more, consider arthrodesis or disarticulation.
- For the treatment of rocker feet, Coleman suggests the following scheme: before 6 years of age, soft tissue treatment, after 6 years of age, subtalar arthrodesis (Grice type), and after 12 years of age, triple arthrodesis.

Clinical case description

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Female patient, 2 years and 10 months old, who attended a pediatric consultation for a routine check-up, with a history of Escobar Syndrome. Born by cesarean section at 35 weeks of gestation, controlled pregnancy, PROM (6 hours), clear amniotic fluid and aspiration of this, dystocic delivery, mild neonatal hypoxia; APGAR 6/7, birth weight: 2200 gr (2.2 Kg), height: 39 cm and WC: 33 cm.

At 3 days of age, she was diagnosed with low-set ears, absence of external auricle, short and asymmetric neck, asymmetric thorax, distal malformations in the limbs, and presence of hyperflexion of the fingers and hip.

Non-exclusive breastfeeding, supplemented with formula, weaning started at 6 months. Low-calorie diet, and physical activity 5 days a week, 1 hour daily. Complete vaccination to date. Neonatal screening performed with normal results.

Figure 1. *Image in right lateral position, where scoliosis and multiple pterygia of the humerus, ulnar and bilateral popliteal fossa are observed.*

Psychomotor Development:

- Head support: 3 months.
- Sitting with support: 6months.
- Side wheel: 5 months.
- He didn't crawl.
- Walked: 2 years.
- Deciduous dentition: 6 months.
- Linguistic stage and psychointellectual development without alterations.

Personal medical history:Mild neonatal hypoxia; ASD (Osteum Secundum) at birth

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measuring 8 mm, with spontaneous closure at 7 months; Cleft Palate at birth, resolved spontaneously at 6 months; Febrile Convulsion at 9 months and Escobar Syndrome.

Figure 2. *Image in anterior and right lateral position, showing camptodactyly, low-set ears and hip dysplasia.*

The family history of pathology highlights:

- Paternal grandmother: Amyloidosis since age 52.
- Paternal aunts:
  - (Figure 3-A): Psychomotor development disorders, pelvic and spinal scoliosis vertebral,episodes of dystonia and muscle contractility. Turner syndrome (ruled out) and Noonan syndrome (under investigation).
  - (Figure 3-B), anatomical deformity in the right clavicle and sacrococcygeal junction.
  - Both paternal aunts have suffered from hyperthyroidism since they were 13 years old.

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Figure 3.*(3-A): Psychomotor development disorders, pelvic and spinal scoliosis vertebral, episodes of dystonia and muscle contractility. Turner syndrome (ruled out) and Noonan syndrome (under investigation).*

(3-B), anatomical deformity in the right clavicle and sacrococcygeal junction.

Currently, the patient's weight is: 9.15 kg, height: 85 cm and PC: 49.5 cm. There is evidence of mandibular micrognathia; scoliosis at the level of the 9th and 12th thoracic vertebrae, with a left concavity and a right convexity, without distal vascular involvement; multiple pterygium in the neck, left axillary fossa, bilateral humero-ulnar joint and bilateral popliteal fossa; localized camptodactyly in the little finger and ring finger of the right hand, hip dysplasia; length asymmetry in the femur (right) and bilateral equinovalgus foot.

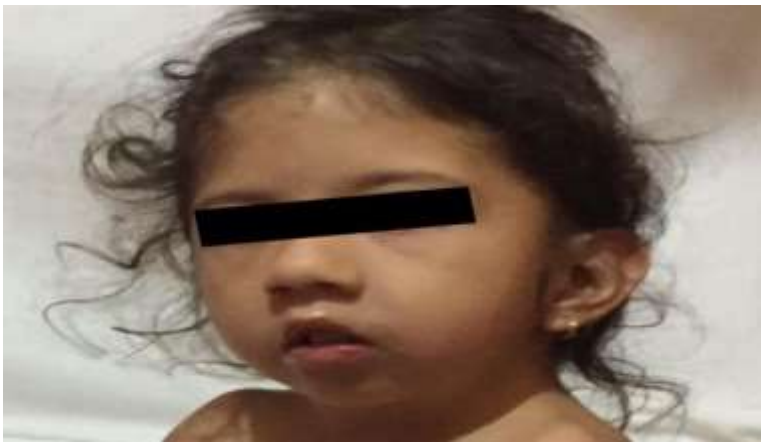

Figure 4.*Micrognathia (abnormally small lower jaw).*

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Figure 5.*Low-set Ears.*

Figure 6.*Scoliosis (right concavity and left thoracolumbar convexity).*

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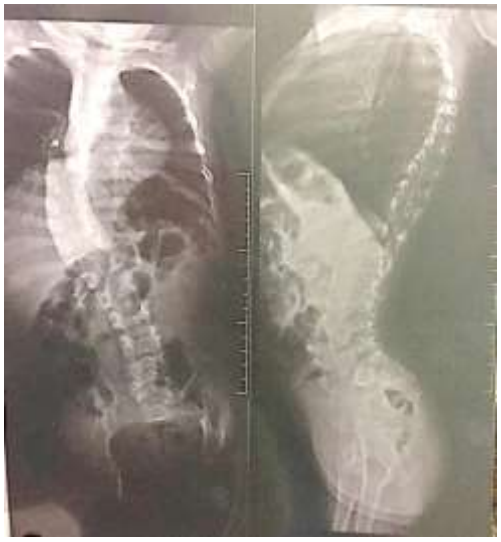

Figure 7. Simple anterior and lateral X-ray showing scoliosis.

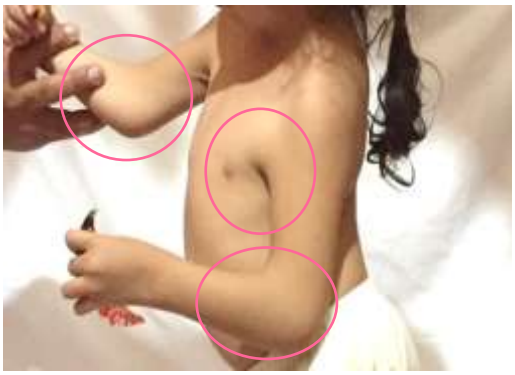

Figure 8. *Pterygium located in the bilateral humeroulnar joint and left axillary fossa.*

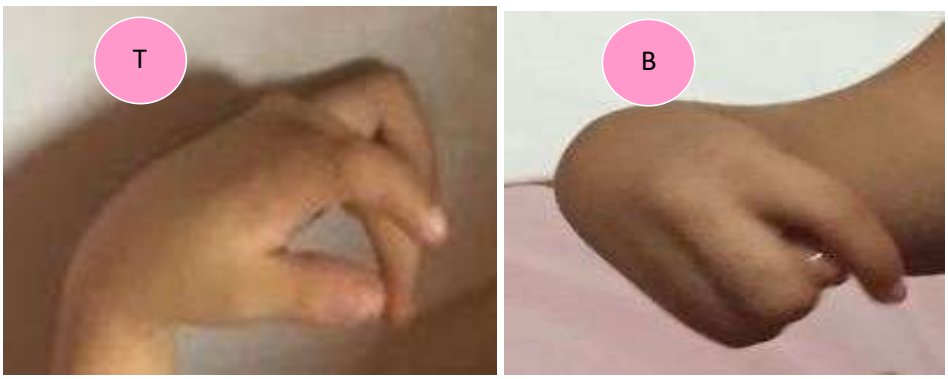

Figure 9.*(TO)Camptodactyly (Left Hand) and (B) Camptodactyly (Right Hand) and limitation of extension and deformity of the interphalangeal joint.*

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Figure 10. Front and back image showing Hip Dysplasia.

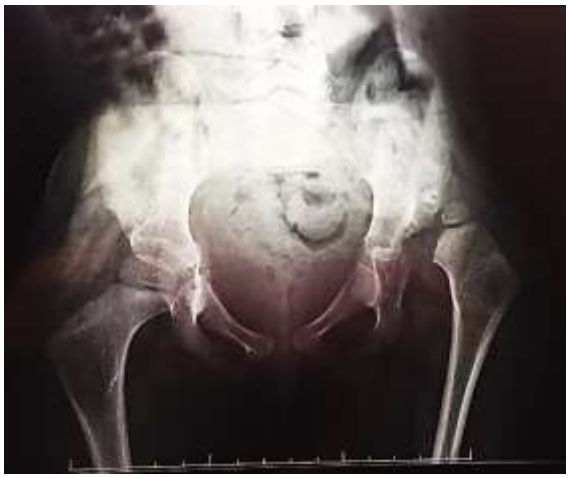

Figure 11. Anterior view radiograph showing dysplasiaHip.

Figure 12. Femur length asymmetry.

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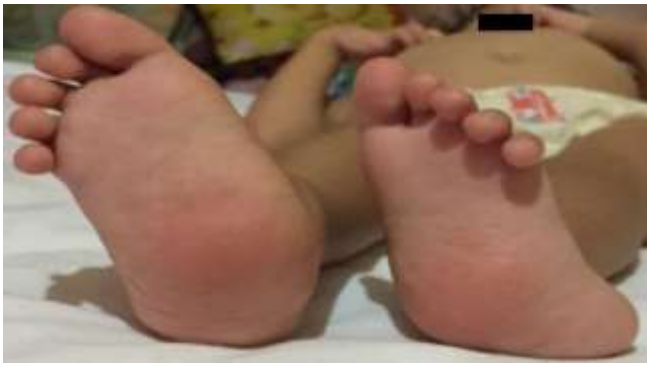

Figure 13. Bilateral equinus valgus foot - Deformity of the foot in which it is permanently in a plantar flexion position..

Figure 14. Radiograph of Bilateral equinus valgus foot.

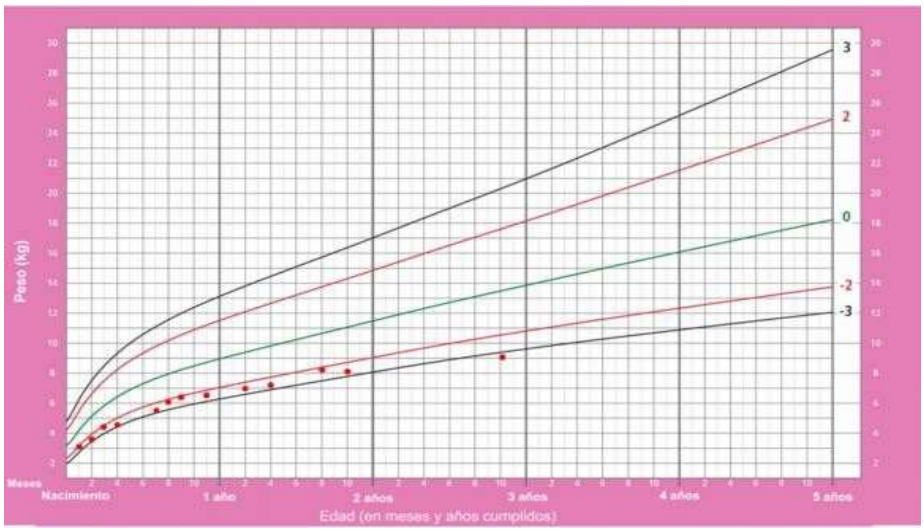

Figure 15. *Weight for age (birth to 5 years).*

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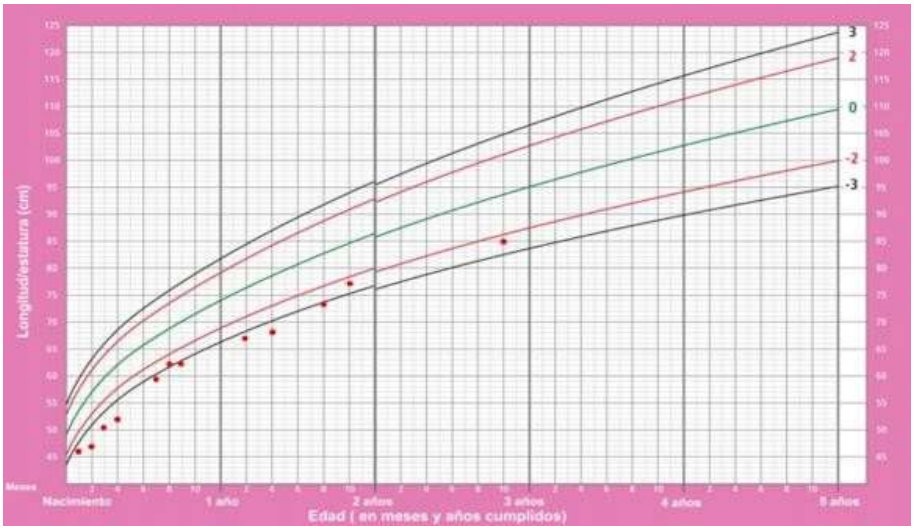

Figure 16. *Size for age (Birth to 5 years).*

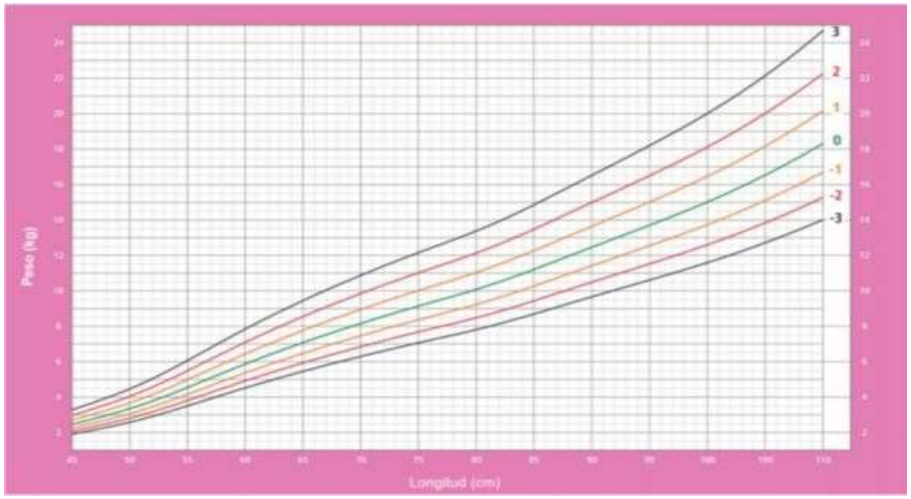

Figure 17. *Weight for length/height (Birth to 5 years).*

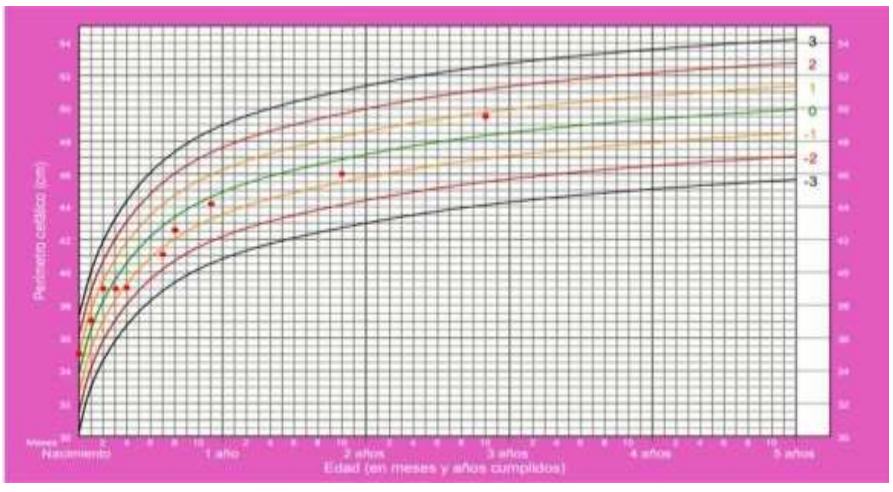

Figure 18. *Head circumference for age. (Birth to 5 years).*

Conclusions

Escobar Syndrome has an unknown etiology, but homozygous or compound heterozygous mutations in the CHRNG gene locus 2q37 and muscle degeneration processes have been associated.

Clinically, it can manifest with particular characteristics and signs in each case.

Epidemiologically, there are 100 reported cases worldwide. In Ecuador, there are no published or evidenced cases.

In the present clinical case, the main alterations are scoliosis, multiple pterygia, camptodactyly, hip dysplasia and bilateral equinovalgus foot, which partially limits the quality of life.

According to WHO growth patterns, it presents:

- Severely underweight.
- Growth retardation.
- Malnutrition.
- He perimeter cephalic HEfind according to age.

The diagnosis is confirmed by genetic testing that shows the CHRNG mutation, however, its absence does not rule out Escobar Syndrome.

The patient does not present pulmonary restriction, despite this being the most frequent complication, an indicator of a good prognosis.

Our case is undergoing karyotype research and corrective surgeries on the hip and feet.

Conflict of interest

The authors of this study declare that they have no actual or potential conflicts of interest with respect to the research, the presentation of results, or the interpretation of the data. No author has received funding, grants, or other forms of support that could influence the objectivity of the research. Furthermore, there are no financial, personal, or professional relationships that could be construed as a conflict of interest in relation to this work. This declaration is provided in order to ensure transparency and integrity in the presentation of the findings of this study and to maintain public confidence in the objectivity of medical research.

Authors' contribution statement

The data collected during the development of this study will be available for review and validation by other researchers and health professionals. Those interested in accessing the data can contact the research team through the contact details provided. This measure seeks to promote transparency and collaboration in scientific research, allowing the verification of results and the expansion of knowledge in the area of study.

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