Nepal Orthopedic Association Journal
https://doi.org/10.59173/noaj.20241001i

Case Report

Non-lethal multiple pterygium syndrome (Escobar syndrome) with bilateral radial club hands: A case report

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ABSTRACT
Escobar Syndrome, the non-lethal variant of multiple pterygium syndrome (MPS), is a rare disorder. It is a subgroup of arthrogryposis multiplex congenita characterized by multiple webbing, fixed joint contractures, and distinctive facies like antimongoloid eyes, ptosis, micrognathia, low-set ears, high-arched palate, down-turned corners of lips, epicanthal folds, and long philtrum. Radial club hand is an uncommon congenital anomaly, and there is a loss of position on the radial aspect of the wrist because of the absence of the radius. This case has a very rare association between Escobar syndrome and bilateral radial club hands, and explores management strategies to improve function.
Keywords: Escobar syndrome; multiple pterygium syndrome; radial club hand

INTRODUCTION
Escobar Syndrome, the non-lethal variant of multiple pterygium syndrome (MPS), is a very rare genetic disorder.[1] The primary manifestations are the presence of multiple pterygium fixed joint contractures with characteristic facies. In lethal MPS, the fetus develops cystic hygroma, arthrogryposis, hydrops fetalis, intestinal malrotation, microcephaly, and congenital diaphragmatic hernia.[2]. The radial club hand is a rare congenital anomaly with deficiency along the radial aspect of the upper extremity. The reported incidence of radial club hand is 1 in 30,000 to 1 in 100,000 live births.[3] This is a very rare case of Escobar syndrome associated with bilateral radial club hand.

CASE REPORT
A 12-year-old presented to us with a chief complaint of deformity of bilateral upper and lower limbs since birth. The deformities were non-progressive. He was unable to walk and extend his elbows. His birth history was unremarkable. There were no peri or postnatal complications, and milestones were normal.
On general examination, the child looked alert and active. He had a dysmorphic look with left-sided ptosis, low sets of ears, prominent epicanthic folds, flat nasal bridge, micrognathia, posterior hairline, and high arched palate. His spine examination revealed mild scoliosis. There were no abnormalities detected on cardiovascular, respiratory, or abdominal examination.

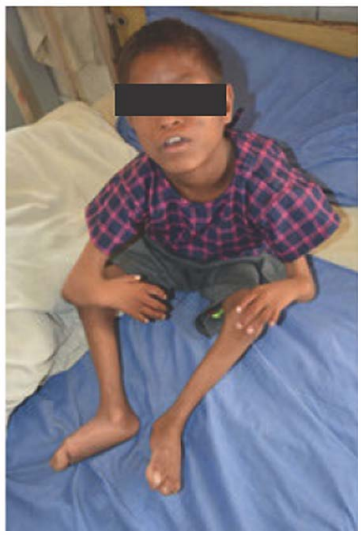

Figure 1. Patient with bilateral elbow and knee pterygia, distinctive facies of Escobar syndrome (dysmorphic face with left-sided ptosis, antimongoloid slants, low sets of ears, prominent epicanthic folds, flat nasal bridge) and bilateral radial club hands with thumb hypoplasia.

Upper limb examination revealed bilateral short limbs with elbow flexion contracture with webs and forearm bowing with radial deviation of wrists. Thumb hypoplasia was present bilaterally with thenar eminence atrophy. There were bilateral elbow flexion contractures with webs. His shoulders and chest were normal. On palpation, both radii were absent. His passive range of movements at shoulder joints was normal. There were flexion contractures of 70 degrees at bilateral elbow joints. There was a restriction in flexion, extension, and ulnar deviation at both wrists and restriction at the IP joint and MCP joint of the thumb bilaterally. All other fingers had a normal range of motion. He could handle most of the objects but with somewhat reduced speed and quality.

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His lower limb examination revealed 70 degrees flexion contracture with popliteal webs. His hip joints and ankle joints were normal, with intact distal neurovascular status

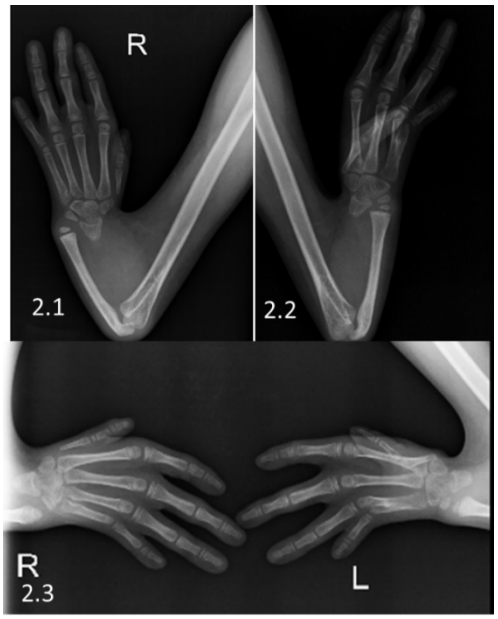

Figure 2.1: Right radial club hand with hypoplastic 1st metacarpal with absent 5th finger phalange.
Figure 2.2: Left radial club hand with hypoplastic 1st metacarpal with bifid 4th metacarpal and absent 5th finger phalange.
Figure 2.3: AP view of bilateral hands

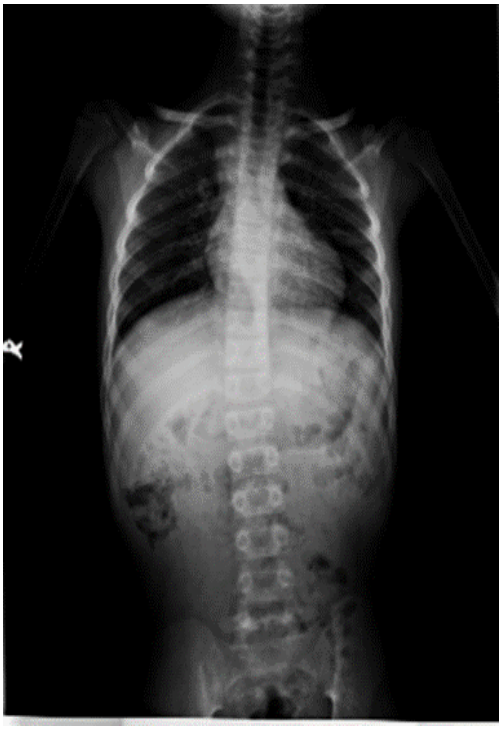

Figure 3: AP view of the spine showing mild right-sided scoliosis

His skeletal survey revealed bilateral absent radii. On the left side, there were only four rays with hypoplastic 1st metacarpal with bifid 4th metacarpal and absent 4th finger phalange. His right hand showed hypoplastic 1st metacarpal with an absent 5th finger phalange. He had a Type 4 bilateral radial club hand according to Bayne and Klug classification. He also had a right-sided mild thoracolumbar scoliosis. The x-rays of lower limbs showed bilateral flexion deformity of knee joints with bilateral patellar hypoplasia with mild tibial bowing.

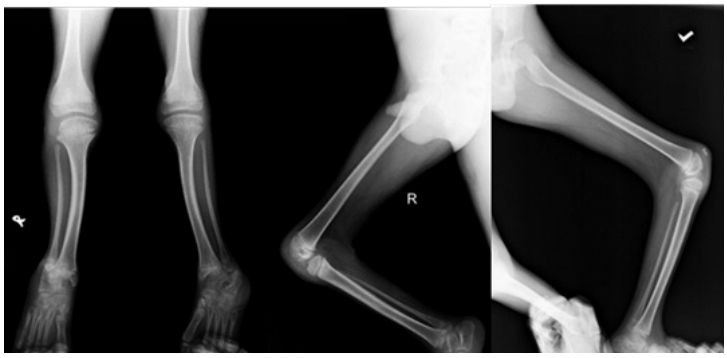

Figure 4: X-rays of bilateral lower extremities showing knee contractures with mild tibial bowing with patellar aplasias

The patient underwent intervention with skeletal traction followed by long-leg stretch casting to correct the bilateral flexion deformity at his knees, which was caused by the pterygium. Given a reasonably good function and adaptation to activities of daily living, it was decided against surgical intervention for the bilateral radial club hands. Post-operatively, complete knee extension was obtained, and an above-knee cast was applied bilaterally. After six weeks, knee orthoses were given.

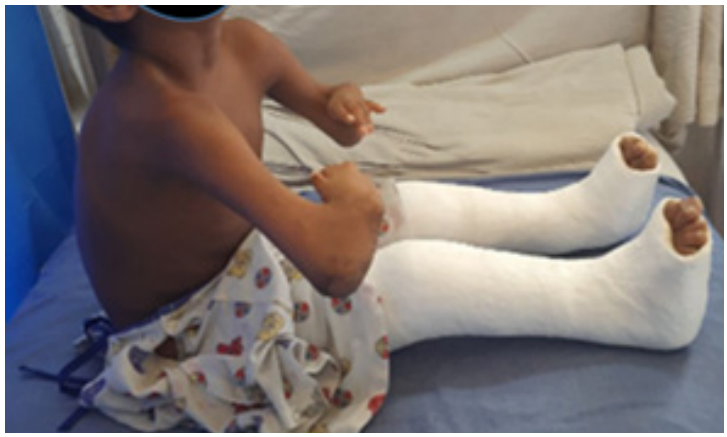

Figure 5: Bilateral knee contractures was corrected after skeletal traction and long leg stretch casting.

DISCUSSION

MPS was first described by J. A. Bussiere in 1902, whereas Escobar *et al*. termed Escobar syndrome for the nonlethal form in 1978.[4]

MPS is a rare genetic congenital disorder characterized by multiple pterygia, multiple joint contractures (arthrogryposis), and skeletal deformities.[1] Our patient had bilateral elbow and knee webbing with flexion contractures with mild right-sided scoliosis. MPS can be divided into two categories, lethal type and nonlethal type - Escobar type. The lethal variant is prenatally diagnosed with ultrasound with cystic hygroma, multiple pterygia, and sometimes stillborn. The Escobar syndrome includes multiple joint contractures with multiple pterygium and distinctive facial features. Our patient falls

under the Escobar variant. The inheritance pattern is usually autosomal recessive. The CHRNG gene mutation has been found to be responsible.[5] Our patient had autosomal recessive, as his parents or sibling had no history of the disease.

Radial deficiencies are the complete or partial absence of the radial border of the upper extremity. Radial club hand is a common radial dysplasia. The reported incidence of radial club hand is 1 in 30,000 to 1 in 100,000 live births. Radial deficiencies may range from hypoplasia of the thumb to the complete absence of radius and first ray. According to Bayne and Klug, congenital radial club hand can be classified into four types, from less severe Type I with defective distal radial epiphysis to the most severe Type IV with a complete absence of the radius, which is also the most common type.[3] Our patient had Type IV bilateral radial club hand with hypoplastic thumbs.

Spranger et al. reported two sisters with Escobar syndrome with severe muscular atrophy.[6] Ezirmik et al. reported a case of Escobar syndrome with patellar aplasia.[7] Our patient also had bilateral patellar aplasia. Karad et al. reported a case of Escobar syndrome with monodactyly.[2] To the best of our knowledge, Escobar syndrome with bilateral radial club hands has not been reported.

In conclusion, we report a case of Escobar syndrome associated with radial club hand, which is extremely rare. In view of the relatively poor results of surgery for the pterygium of the knee, skeletal traction and serial casting were chosen as treatment, and this resulted in a straight knee, allowing upright ambulation. There was a trade-off between function and cosmesis for the radial club hands, so in light of good functional adaptation, no intervention was recommended for the upper extremity.

CONFLICT OF INTEREST

None

CONSENT

Written informed consent was obtained from the patient.

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