THE LIMPING CHILD: A RARE CASE OF MULTIPLE PTERYGIUM SYNDROME, ESCOBAR TYPE
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INTRODUCTION

Limping is a common problem in children and adolescents. The differential diagnosis is extensive which depends on the child’s age, symptoms, onset and duration of the symptoms. Rarely, limping can be attributed to a genetic disorder such as Multiple Pterygium (Escobar) syndrome that presents with multiple congenital anomalies and multi-system involvement.

CASE REPORT

Patient is a 12-year-old boy who was born with lower limb deformities of both legs. At 4 months, he was unable to extend his right elbow. As the patient grew, there was delay in standing. At 3 years, he started to learn how to stand and walk but with a limping gait. Both ankles were hyperextended and externally rotated with limitation of movement. His right elbow joint had only 45-90° range of motion. At present, the patient appears thin and has short stature. He can walk but with a limp and difficulty in balancing. There is external rotation of the lower extremities at hip joint and plantar flexion with contractures at both ankle joints. There is also a deformity with limitation of movement of the right elbow with excess soft tissue fold. The left leg appears longer than the right. No sensory deficits or mental abnormality were noted.

IMAGING FINDINGS

Skeletal Survey was done. AP and lateral view of the lumbar sacral spine shows spina bifida occulta at the 5th lumbar and 1st sacral vertebrae (Fig 1 A and B). Lateral views of the forearms reveal proximal radial dislocation with no evidence of fracture on the left (Fig 2 A) and skin contracture (pterygium) at the antecubital area on the right (Fig 2 B) as also seen on the right elbow joint (Fig 2 C). A photograph of the antecubital excess soft tissue on the right is also shown (Fig 3) The AP (Fig 4 A) and Frog-leg (Fig 4 B) radiographs of the hip demonstrate congenital hip dislocation, bilaterally. Both fibula are absent as shown in Fig 5 A and B. AP view of both feet shows persistent plantar flexion (Fig 6 A and B) with clubfoot deformity on both sides (Fig 7 A and B).

DISCUSSION

The clinical manifestations and pattern of external malformations seen in the patient closely approximates that of the Multiple Pterygium (Escobar type) Syndrome, a rare inherited disorder manifested by growth retardation, facial and genital anomalies, and widespread musculoskeletal deformities. The autosomal dominant type is characterized by multiple pterygium with or without mental retardation. The autosomal recessive type is characterized by multiple joint contractures with marked pterygia, dysmorphic facies and cervical vertebral anomalies. Pterygia-cutaneous webbing usually associated with flexion joint contractures are the predominant feature of the syndrome. Patients may manifest with short stature, abnormal facies and webbing of the neck, antecedital, digital, popliteal, and intercrural areas.

Features of Escobar Syndrome * (those in bold letters are manifested by the patient)

Head: Neck, antecubital, digital, popliteal, intercrural, and antecubital pterygium
Resemblance: Congenital hip dislocation, dislocation of the radial head, flexion deformity of the fingers and thumbs, talipes calcaneovalgus, talipes equinovarus, and rocker-bottom feet
Small and much small mandible with pointted receding chin, long philtrum, synophrys, low-set ears, epicanthal folds, down-slanting palpebral fissures, blepharophimosis, eye puffiness in some cases, down-turned angles of the mouth, lip pits, occasional clavical palate, and squint-like shape of the tongue (lingua coelicares)
Spinal Fusion of the cervical vertebrae, hypoplasia of spinous process, spina bifida occulta
Thorax: Small heart, lung hypoplasia
Genital: Absence of the appendix, atresia of the colon
Growth and development: Growth and occasional mental retardation
Behavior and performance: Peculiar unsteady gait with crouched stance and conductive deafness

CONCLUSION

A child’s first steps in life are wondertom to the parents and the beginning of new discoveries for the child. The differential diagnoses of a limp in a child are broad. Normal age-related gait must first be differentiated from gait impairment. Once gait impairment is identified, differential diagnoses are narrowed as the history and physical examination findings begin to suggest a source of the limp. In many instances, a diagnosis is reached with the assistance radiographic findings, initially by plain radiographs.

Escobar syndrome, although rare, should be considered in this case. Aside from the fact that the patient presents with musculoskeletal abnormalities that are suggestive of this syndrome, the radiographic findings have been helpful in this case. Further imaging studies should be contemplated to have more documentation of this syndrome such as MR Imaging, which is most likely the imaging of choice when evaluating children who are being considered for surgical resection of the pterygium or correction of contractures.

REFERENCES AND RELATED ARTICLES

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