Case Report — The Eurasian Journal of Medicine — EAJM 2012; 44: 117-21

Escobar Syndrome Mimicing Congenital Patellar Syndrome

Konjenital Patella Sendromunu Taklit Eden Escobar Sendromu

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Abstract

Multiple pterygium syndrome (MPS) is a syndrome that is characterized abnormal face, short length and skin pterygiums on some body legions (servical, antecubital, popliteal, interdigital and on neck). It is also called as Pterygium Colli syndrome, Escobar syndrome or Pterygium syndrome. Escobar (multyple pterygium) syndrome is a rare syndrome. Intrauterin growth reterdation, abnormal face, wide-spead pterygiums that resulted in joint contractures, ptosis, chryptoorchidism, patellar dysplasia and foot deformities are seen on this syndrome. Primarly autosomal resesive crossing are observed; also autosomal dominant and X-linked crossing. This case were presented as it has components of Escobar syndrome and Isolated Patellar Aplasia syndrome in same time.

Key Words: Escobar syndrome, Isolated Patellar Aplasia syndrome, Pterygium syndrome

Özet

Multipl Pterjium Sendromu anormal yüz, kısa boy, vücudun bazı yerlerinde (servikal, antekübital, popliteal, parmaklararası) deri katlantıları ile karakterize bir sendromdur. Ayrıca Pterjium Kolli Sendromu, Escobar Sendromu veya Pterjium Sendromu diye adlandırılır. Escobar (Multipl Pterjium) Sendromu nadir bir sendromdur. Bu sendromda intrauterin gelişme geriliği, anaormal yüz, kontraktürler sonuçlanan geniş-yayılımlı pterjiumlar, pitosis, kriptoorşidizm, patellar displazi ve ayak deformiteleri görülür. Primer olarak otozomal resesif geçiş olmak üzere otozomal dominant ve X kromozomuna bağlı kalıtım görülür. Bu vaka Escobar ve Konjenital Patellar Sendromun komponentlerine aynı anda sahip olduğu için sunulmaktadır.

Anahtar Kelimeler: Escobar sendromu, İzole Patellar Aplazi sendromu, Pterygium sendromu

Introduction

Multiple pterygium syndrome (MPS) is characterized by an abnormal face, short height and skin pterygiums on certain body regions (e.g., cervical, antecubital, popliteal, interdigital) and on the neck. MPS is also called pterygium colli syndrome, Escobar syndrome and pterygium syndrome [1].

Escobar (multiple pterygium) is a rare syndrome that presents with intrauterine growth restriction, abnormal facial features, widespread pterygiums that result in joint contractures, ptosis, cryptorchidism, patellar dysplasia and foot deformities are observed in this syndrome. Inguinal hernias and cranial ventriculomegaly have also been established as components of Escobar syndrome [2].

The most common element of this syndrome is joint contracture, followed by scoliosis, thoracic deformity, rocker bottom foot, syndactyly, camptodactyly, cleft palate, and abnormal facial features [3]. Primary autosomal recessive crossings are observed, as are autosomal dominant and X-linked crossings [4]. The present case has components of both Escobar syndrome and isolated patellar aplasia syndrome.

Case Report

This patient is an 8-year-old female. Her delivery was normal. At birth, upon examination the only abnormality reported was an abnormal face.After a few days, the patient and her mother were discharged from the hospital as usual. The patient's family included 5 children (3 girls, 2 boys). Her brothers and sisters had normal physical examination findings. There was no recall of drug use, radiation exposure, or serious chronic illness of the mother during pregnancy. There was no kinship between the patients' parents. This syndrome was not detected in any other family members.

At birth, her weight was 2635 g, length was 47 cm, and caudal diameter was 24 cm. There were no pathological heart sounds on cardiac examination. The patient had bilateral patellar aplasia (Figures 1-4). The findings of other examinations included the following: micrognathia; a high palate; large low-set ears; a short neck; antecubital, popliteal, and cervical skin folds; scoliosis; flexion contractures of the knee and elbow; total lack of hand extensor tendons (Figures 5-8). Scoliosis was not severe (Figure 9). Echocardiography and abdominal ultrasonography were normal. The karyotype was 46, XX.

Received: August 11, 2011 / **Accepted:** September 07, 2011

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doi:10.5152/eajm.2012.26

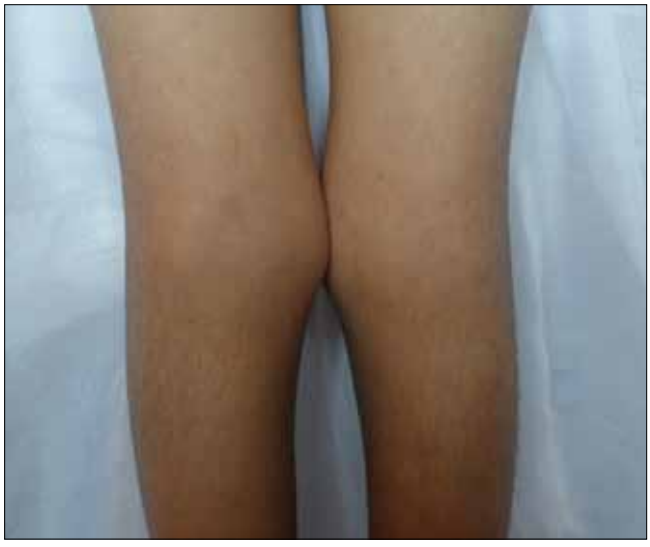

Figure 1. *Bilateral patellar aplasia in clinical examination (AP).*

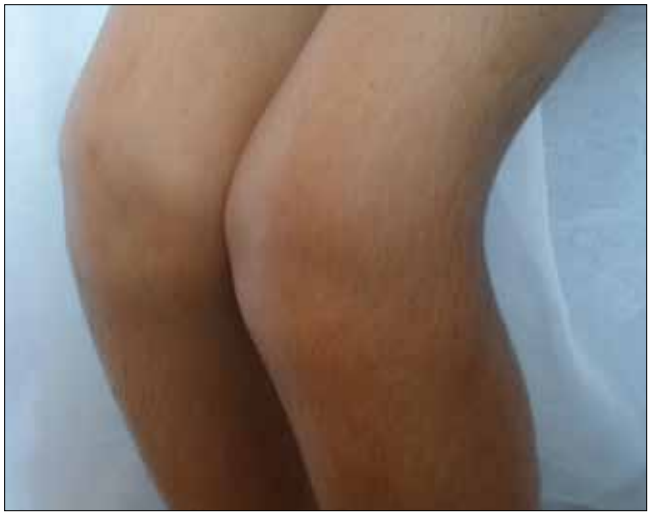

Figure 2. *Bilateral patellar aplasia on clinical examination (lateral).*

On x-ray there was bilateral patellar aplasia (Figures 1, 10, 11). Bilateral patellar aplasia was also detected by magnetic resonance imaging (Figures 12-14). With regard to the laboratory findings, CBC and biochemical panel were normal. Thyroid hormones were in the normal ranges. An additional systemic workup was negative.

Discussion

Multiple pterygium syndrome (MPS) is also known as Escobar syndrome, pterygium colli syndrome or, simply, pterygium syndrome [5]. Multiple pterygium (Escobar) syndrome is a rare, autosomal recessive inherited disorder manifested by growth retardation, facial and genital anomalies, and widespread musculoskeletal deformities. Pterygia-cutaneous webbing, usually

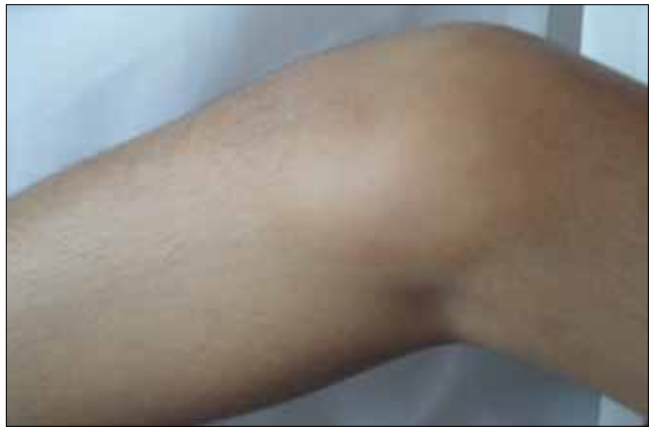

Figure 3. *Right knee.*

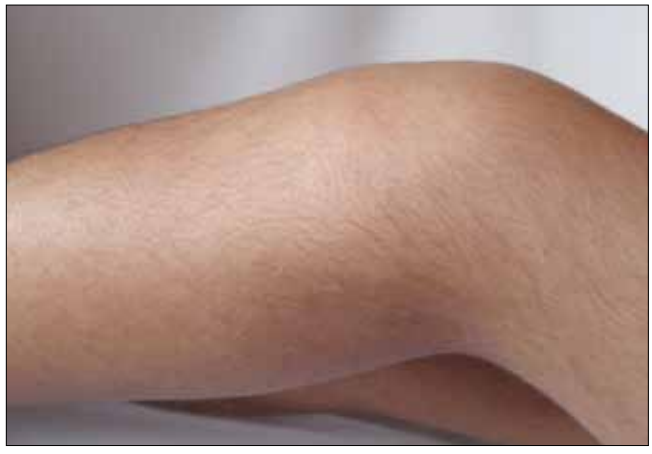

Figure 4. *Left knee.*

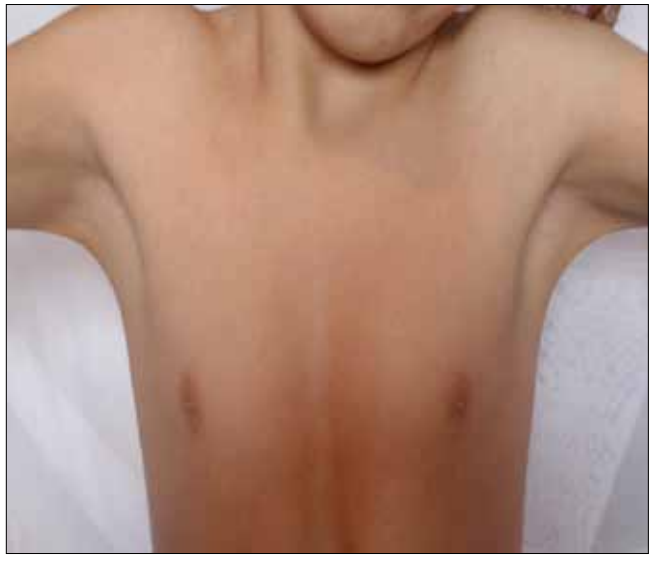

Figure 5. *Bilateral axillar pterygiums.*

Figure 6. *Lack of extensor tendon.*

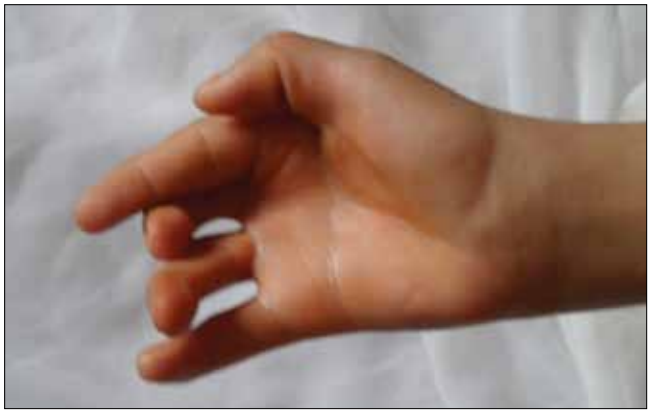

Figure 7. *Flexion posture of hand.*

Figure 8. *Low neck-hair border.*

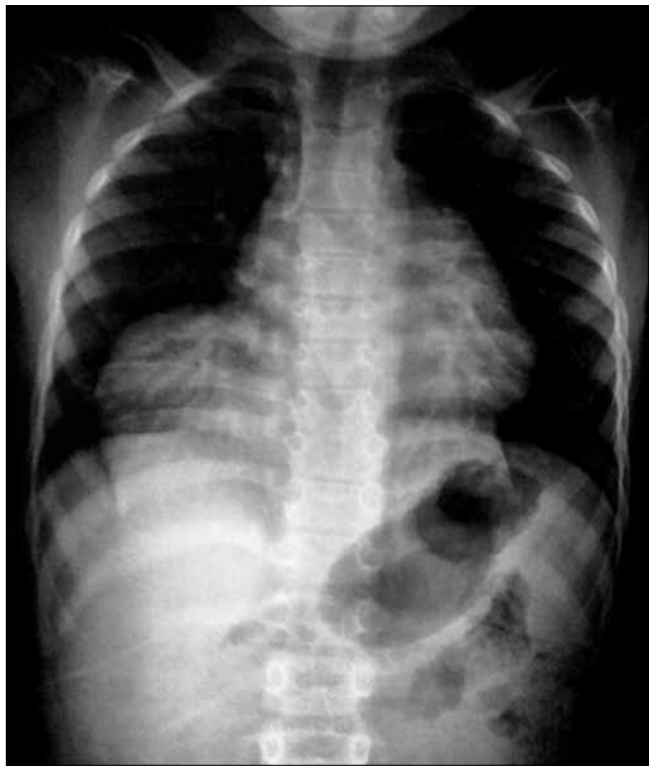

Figure 9. *PA lung graphy.*

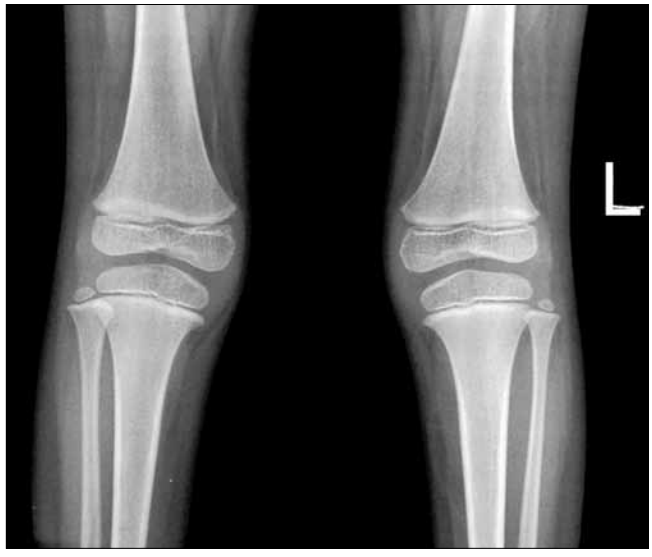

Figure 10. *AP bilateral knee.*

associated with joint contractures, is the predominant feature of the syndrome [6]. Escobar syndrome was first defined in 1902 by Bussiere et al. [7], and later revised in 1978 by Escobar et al. [8]. Approximately 50 cases were presented. Generally, this syndrome has autosomal recessive inheritance [9].

This syndrome presents with growth retardation and generally small body size, as observed in our patient. Additional possible features include antimongoloid palpebral fissures and pytosis, innerly folded contuses, hypertelorism, micrognathia, a long philtrum, cleft palate, an ugly and board-flat

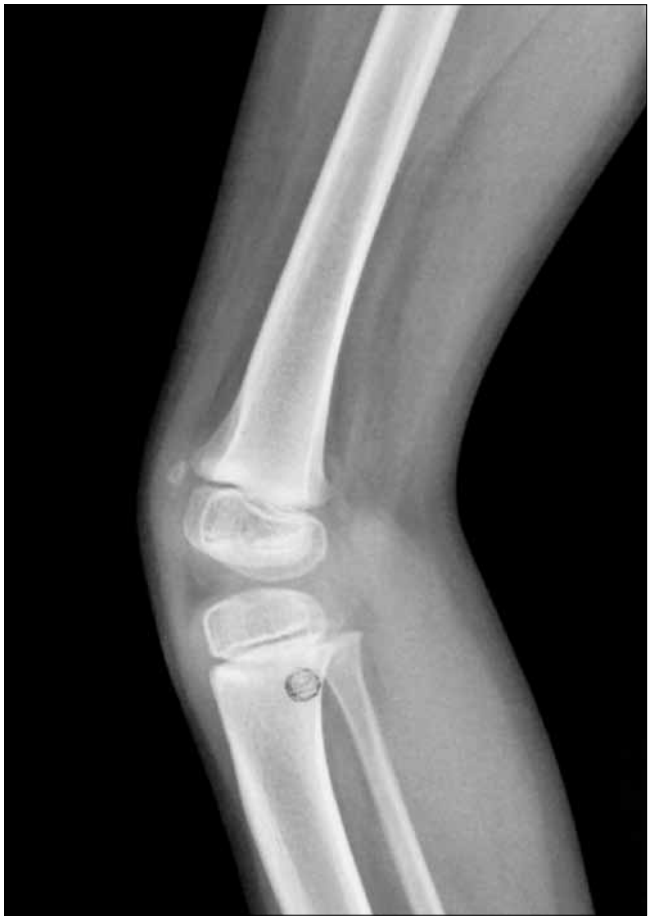

Figure 11. *Right knee.*

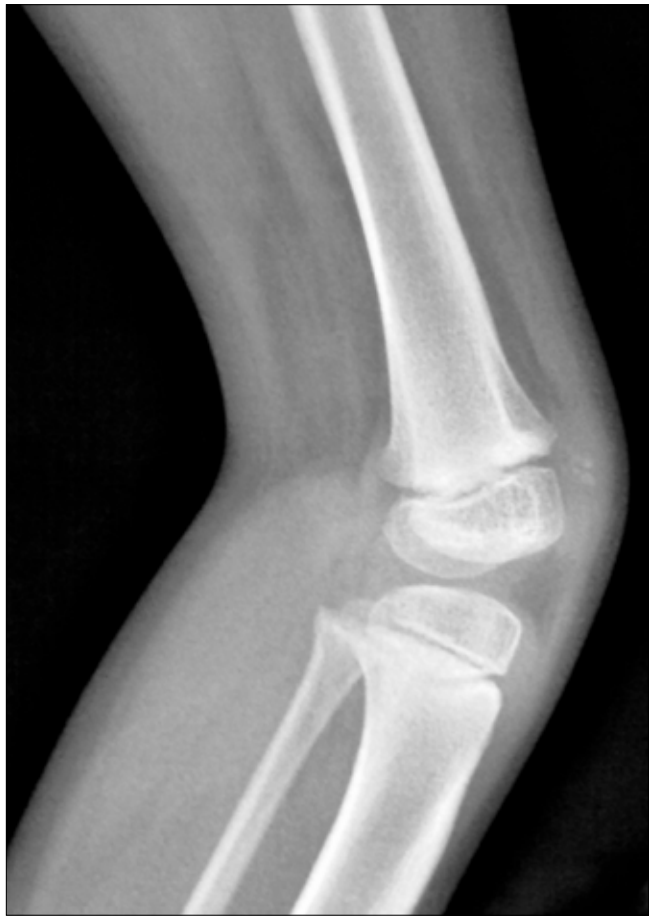

Figure 12. *Left knee.*

face, and low-set ears [9]. A dysmorphic face contains palpebral adhesions, a small mouth, a flat and short nose, cleft palate, micrognathia and ear abnormalities [10]. A short neck and cervical skin folds have also been characterized in this syndrome [1]. Musculoskeletal abnormalities including scoliosis, hypotonia, and lower limb deformities are also common [11]. Pathologic changes can also result in neuromuscular pathology [12]. General abnormalities may be identified, but on long-term follow-up secondary sexual changes are generally in the normal range [13].

This syndrome is autosomal recessive, but other inheritance patterns can also be observed [1]. Goh et al. [14] reported the case of a 4-year-old boy who was short and had pterygiums in his axillary and antecubital areas. McKeown and Haris reported autosomal dominant inheritance in three children with a mother affected by MPS [15]. Carnevale et al. [16] reported X-linked dominant inheritance in three cases over a period of seven decades. All girls of affected fathers developed MPS, but no boys developed MPS [16].

Wide-spread pterygiums attract clinical attention in Escobar syndrome and include antecubital, axillary, cervical, popliteal and intercrural legions. Camptodactyly, syndactyly, equinovarus and rocker-bottom feet can also contribute to these patients' clinical appearance. Cryptorchidism or lack of the labia majoris can also be detected. Additional clinical features include patellar aplasia or dysplasia, scoliosis, kyphosis, vertebral fusions, and costal abnormalities [9].

Additional defects that can be detected as part of this syndrome include the following: long-shaped vertebral bodies with clefts, a loosening of the of antero-posterior diameter, fusion deficiency of the posterior neural arc, costal fusions, long clavicles with lateral hooks, dislocation of the radial head, distal radio-ulnar separation, muscle atrophy, dislocations of the hips, hypoplastic and/or wide nipples, loss of hearing, diaphragmatic hernia, hypospadia and heart defects. Many patients with this syndrome are ambulatory. Intelligence is generally normal. Kyphoscoliosis, episodes of apnea and/or dyspnea due to a small chest, and pneumonia (50% of these patients die of pneumonia) can also be encountered. Pterygiums can be present at the time of diagnosis and can result in contractures [1].

Treatment must be targeted at resolving the specific complaints and needs of patients. Early and effective physical exercises must be implemented. Most suitable orthopedic surgeries

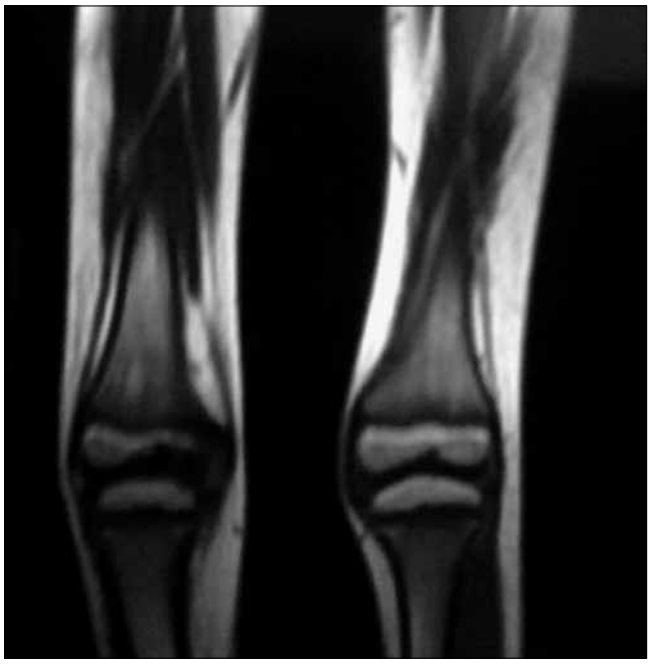

Figure 13. *Bilateral knee on MRI.*

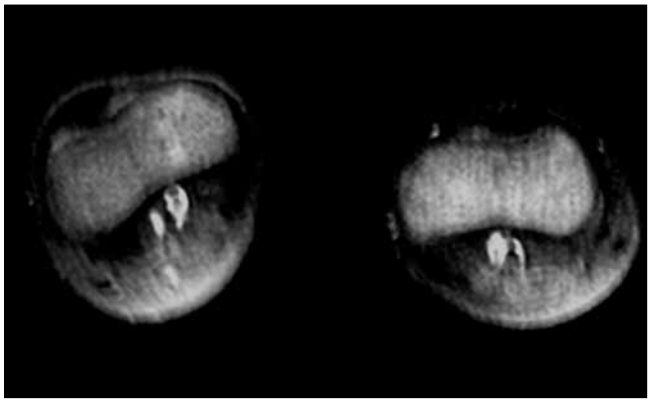

Figure 14. *Sagittal MRI.*

must be applied in the light of clinical evidence and according to specific needs. These procedures include surgical fusion for scoliosis, release approaches for hand and foot abnormalities, and other surgical procedures for other specific components [9].

In conclusion, Escobar syndrome and congenital patellar syndrome are two different syndromes that include patellar aplasia as one component of their presentation. Patellar aplastic syndromes are a broad group of syndromes that affect the entire body. Even if this case had a normal karyotype, multiple pterygiums and some components of Escobar syndrome, it would be mimicking isolated congenital patellar aplasia (congenital patellar syndrome).

In the medical literature, Escobar syndrome has presented with many different clinical manifestations. However, patellar aplasia and multiple pterygiums without other clinical findings have never been reported in the same case. In conclusion, we identified a case presenting as Escobar syndrome mimicking congenital patellar syndrome.

Conflict of interest statement: The authors declare that they have no conflict of interest to the publication of this article.

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