



## Escobar (multiple pterygium) syndrome: Multidisciplinary approach to a very rare syndrome

Escobar (multiple pterygium) sendromu:  
Çok nadir bir sendroma multidisipliner yaklaşım

Murat Celal Sözbilen, MD.,<sup>1</sup> Hüseyin Günay, MD.,<sup>1</sup> Funda Atamaz Çalıř, MD.<sup>2</sup>

<sup>1</sup>Department of Orthopedics and Traumatology, Medical Faculty of Ege University, İzmir, Turkey

<sup>2</sup>Department of Physical Medicine and Rehabilitation, Medical Faculty of Ege University, İzmir, Turkey

### ABSTRACT

Escobar (multiple pterygium) syndrome is progressive and a very rare syndrome. It is generally characterized by multiple pterygia that are found in the cervical, antecubital and popliteal regions. In this report, we present the treatment management of a 14-year-old case with late stage Escobar syndrome who was admitted due to multiple pterygia and bilateral knee contractures. The treatment of this case was conducted in collaboration with the department of orthopedics and the department of physical therapy. The treatment of the case was done in three stages: soft tissue procedures, femoral recurvatum osteotomy and triple arthrodesis, respectively. In addition to all these, the patient did intensive physical therapy for one year. As a result of the operations and physical therapy, adequate muscle strength was obtained.

**Keywords:** Escobar syndrome; femoral osteotomy; multiple pterygium syndrome; rehabilitation.

### ÖZ

Escobar (multiple pterygium) sendromu progresif ve oldukça nadir görülen bir sendromdur. Genellikle servikal, antekübital ve popliteal bölgede multiple pterygiumlar ile karakterize edilir. Bu yazıda, 14 yaşında multiple pterygium ve iki taraflı diz kontraktürleri olan Escobar sendromlu geç dönem bir olgunun tedavi yönetimi sunuldu. Bu olgunun tedavisi Ortopedi Anabilim Dalı ve Fizik Tedavi Anabilim Dalı bölümlerinin işbirliği ile yürütüldü. Hastanın tedavisi sırasıyla, yumuşak doku işlemi, femoral rekurvatum osteotomisi ve üçlü artrodez olmak üzere üç aşamada yapıldı. Tüm bunlara ek olarak, bir yıl boyunca yoğun fizik tedavi uygulandı. Uygulanan ameliyatlar ve fizik tedavi sonucunda, yeterli kas gücü sağlandı.

**Anahtar sözcükler:** Escobar sendromu; femoral osteotomi; multiple pterygium sendromu; rehabilitasyon.

Escobar syndrome is generally a progressive syndrome with autosomal recessive inheritance. It is characterized by multiple pterygia (contracture band on the flexor side of joints) that are found in the cervical, antecubital and popliteal regions.<sup>[1]</sup> Although the inheritance of the disease was reported as autosomal recessive, autosomal dominant and X-linked lethal multiple pterygium syndromes have also been reported in the literature. However, the gene locus of Escobar syndrome was not determined yet.<sup>[1-2]</sup> It is one of the rarely seen diseases.<sup>[3]</sup> The other hallmarks of this syndrome are typical facial features such as cleft palate, low-set ears, maloccluded

teeth, micrognathia, ankyloglossia and skin folds in the neck. This syndrome can also manifest itself with kyphoscoliosis, pes equinovarus, syndactyly, camptodactyly or clubfoot (rocker bottom) and the mental functioning of patients are usually normal. Besides, joint contractures, muscle weakness and kyphoscoliosis lead to restrictive decline in pulmonary function in most patients; thus the ambulation is restricted.<sup>[4]</sup>

We report a case with Escobar syndrome who was admitted due to multiple pterygia and bilateral knee contractures, which is very rare. He has a normal

intellectual development, joint contractures, hand-face anomalies, pectus excavatum deformity and had been ambulated with wheelchair before treatment.

### CASE REPORT

A 14-year-old male patient was admitted to the department of orthopedics with difficulty in walking due to multiple pterygia and bilateral knee contractures. His parents have a consanguineous marriage and one of his brothers was born with anatomical anomalies, who died due to respiratory distress on the second day of his life. Our patient had not received any medical or surgical treatment before he was admitted to our hospital. The patient and his legal guardian provided signed informed consent for publication of the images and details of the case in *Eklem Hastalik Cerrahisi*.

The physical examination of the patient revealed that cranium was scaphocephalic with low-set ears; we also observed low hairline and extensor pollicis longus hypoplasia, multiple joint contractures, micrognathia and vertebral anomalies. The neck extension was limited. In C3-7 block, vertebra was determined on the direct radiographies in which scoliosis was observed in the thoracic region with opening facing left and in the thoracolumbar region facing right. T3-6 vertebral spaces appeared to be fused and kyphosis was present (Figure 1). There were flexion deformities and webbing across all flexion creases in the legs. There was



**Figure 1.** Multiple pterygia at physical examination.

bilateral pes equinovarus, clubfoot and syndactyly of the second and third toe. High degree muscle, tendon atrophy and 2/5 muscle strength were observed in his lower extremities. Although his respiratory effort was limited, he did not have major systemic disorder except for musculoskeletal anomalies. On the other hand, no mental retardation and genital system anomalies were observed. The aforementioned symptoms were taken into consideration for further treatment planning. The triple surgery series included the bilateral hamstring muscle-tendon release, biceps femoris muscle lengthening and distal femoral recurvatum osteotomy, respectively.

For surgical technique, in the first session in prone position, the skin was incised on popliteal fossa from middle line of femur to cruris with single-level skin Z-plasty in both knees. The sciatic nerve and popliteal artery were dissected; thereafter Z-plasty lengthening was applied to hamstring and biceps femoris muscle groups. Medial and lateral posterior capsulotomy were performed. Besides, the popliteal artery was taken into consideration during dissection. At the postoperative examination, there was lack of extension, therefore serial casting has been supplemented and changed in two-week intervals for eight weeks. At the end of this process, the patient was transferred to a physical rehabilitation unit.

At physical therapy, both passive and active stretching exercises were performed in string for a month, five days per week. Passive stretching prone position was initiated at least for half an hour, ever-increasingly. Because of neuropathic pain in extremities and lumbar region, pregabalin was started at 75 mg twice daily with 30 min TENS/day that was applied for analgesia. Prior to treatment, the pain severity was determined as 100 mm in visual analog scale (VAS) but it was reduced to 20 mm at the end of the first month. In the meanwhile, lumbar, quadriceps and gluteal isometric exercises were applied. Breathing exercises were initiated and the increase in cardiovascular endurance with arm ergometer exercises was provided. At the end of the first month, the lack of extension was 30 degrees less than those at the beginning of treatment; hence the patient was supported with long leg braces and walker. Lower limb stretching and strengthening, trunk muscle strengthening exercises, treadmill and bicycle exercises were given as home exercise program.

The second surgery was carried out six months after the first operation. The operation in supine position was performed through a lateral skin incision that was extended by 8 cm proximal to the lateral epicondyle of the femur without use of

a tourniquet. The vastus lateralis muscle was freed from the intermuscular septum. The periosteum was incised longitudinally and retracted with retractors to expose the cortex at the planned level of the osteotomy. After recurvatum osteotomy, 3.5 mm wide locking compression plate was placed by the angle of the knee at full extension (Figure 2). Bilateral long leg cast were applied during six weeks at full extension. After six weeks, lack of extension was not observed. Therefore, active, passive and isometric exercises were initialized immediately. When bone healing was observed, the patient was ambulated with full weight bearing with locked hip-knee-ankle-foot orthosis (HKAFO) in the eighth week. The patient had difficulty using long leg orthosis due to the foot deformities. As a result of that, the patient underwent the third session surgery of bilateral achilloplasty, crescentic calcaneal osteotomy and triple arthrodesis at the third month of rehabilitation. During the sixth week, short leg cast was applied and then the patient was transferred to the rehabilitation clinic in order to adapt to the use of the orthosis. Foot-ankle and

knee active-passive strengthening exercises, trunk muscle strengthening exercises, postural control and weight bearing were practiced. In addition to this, the patient continued to perform arm ergometer exercises, treadmill and bicycle exercises in order to increase the cardiovascular endurance. The patient received a home exercise program and was discharged from the hospital (Figure 3).

## DISCUSSION

Multiple pterygium syndrome is characterized by flexion contractures at birth. In the literature, 50 Escobar syndrome cases have been reported but there are only a few case reports in the literature that describe the surgical treatments for Escobar syndrome.<sup>[4,5]</sup> Although the results of surgical approaches for anomalies of neck zone and vertebral were described well, sufficient data is unavailable in the literature about surgical orthopedics approaches for anomalies in lower extremity with rehabilitation programs that were applied after the surgery.<sup>[4]</sup>

The largest series were reported by Joo et al.,<sup>[6]</sup> that described the prevalence of vertebral anomalies and surgical approach but not enough data was presented about lower extremity surgical approaches or their results. Bellamy et al.<sup>[7]</sup> evaluated rehabilitation results of knee flexion contractures in seven cases without mentioning the surgical results. In the study



**Figure 2.** (a) Preoperative left femur. (b) After recurvatum osteotomy postoperative first year. (c, d) Postoperative second year: anteroposterior and lateral X-rays.



**Figure 3.** Postoperative second year: patient can stand and walk with a walker.

of Song et al.,<sup>[8]</sup> an antecubital pterygium case was reported which was treated with Ilizarov technique and resulted in short-term recurrence. Besides, surgery results of lower extremity deformities in seven case series that were applied with soft tissue procedures including Z-plasty, hamstring muscle release, fibrous band release and capsulotomy were given in the study of McCall and Budden.<sup>[9]</sup> As a consequence, the knee arthrodesis had to be applied due to inadequate correction. In addition, the encountered problems under the regional anesthesia may be due to advanced vertebral deformities, while under general anesthesia the problems may have resulted from difficulties in intubation and ventilation.<sup>[10]</sup>

In our case, lack of extension was reduced by 30 degrees after soft tissue procedures and full extension was obtained by femoral recurvatum osteotomy. After soft tissue release and lengthening operations that were combined with femoral and calcaneal osteotomies, adequate muscle strength was obtained. Enhanced muscle strength can provide a range of motion that is close to the normal range of motion; thus daily activities could be done independently. Prior to the surgeries, the patient was able to sit without support. But after surgery the patient also gained the ability to walk with the support of orthosis and a walker. These results demonstrate that the orthopedic interventions and intensive physical therapy were adequate and successful.<sup>[11,12]</sup>

Review of literature confirms that there are few documented cases or case series about the Escobar syndrome. This case demonstrates a 14-year-old male patient with multiple pterygium syndrome treated at a late stage. The patient presented in this case had never been able to stand up and had been confined to wheelchair ambulation until he was 14 years old. At the end of a one-year program of orthopedic surgery and rehabilitation, the patient is now able to walk and do most of his daily routine independently.

It is noteworthy that the contractions recur in early postoperative period in studies which only soft tissue release procedures are applied, despite the intense physical therapy. The osteotomy is considered as a necessity in order to use long leg orthosis and ease of initial stage of physical therapy. With the reason of better gait and posture, adjunctive procedures are deemed to be necessary including triple arthrodesis and calcaneal osteotomy for foot deformities.

As a conclusion, in addition to soft tissue procedures in multiple pterygium syndrome, more studies are needed about the operation of femoral correction and its rehabilitation in terms of comparison with data from the literature. This is because osteotomy surgery and its rehabilitation will gain much more importance in the future for patients with this syndrome.

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