Escobar Syndrome (Multiple Pterygium Syndrome) Associated with Thoracic Kyphoscoliosis, Lordoscoliosis, and Severe Restrictive Lung Disease: A Case Report

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Abstract *Background:* Escobar syndrome or multiple pterygium syndrome is characterized by a web across every flexion crease in the extremities, most notably the popliteal space. In addition, this syndrome is associated with two other structural anomalies: a vertical talus and congenital lordoscoliosis. We present a case report of a patient with Escobar syndrome who was initially managed conservatively and subsequently had severe and debilitating progression and respiratory decompensation ultimately requiring surgical intervention. *Study Design:* Case report. *Methods:* After preoperative evaluation by a pediatrician, pulmonologist, and otolaryngologist, the patient underwent one-stage anterior and posterior spinal fusion with instrumentation as well as multiple osteotomies, rib resections, and vertebrectomies. *Results:* The patient's postoperative course was complicated by wound necrosis requiring irrigation and debridement, a urinary tract infection, and a tracheostomy for persistent atelectasis. The patient eventually recovered from all complications. There were never any focal neurologic deficits. The patient had a 3-year follow-up with radiographically confirmed maintenance of correction. Fusion was obtained in the anterior and posterior segments. Clinically, the patient is able to stand upright, can participate in functional activities, and has not required any pain medication. The patient's functional vital capacity improved from 23% predicted preoperatively to 60% predicted postoperatively. *Conclusions:* Patients with severe spinal deformity secondary to Escobar syndrome can be successfully treated surgically. We propose early surgical intervention in this group to prevent curve progression, restrictive lung disease, and the need for complex salvage procedures.

Key words Escobar syndrome · spinal deformity · restrictive lung disease · multiple pterygium syndrome · lordoscoliosis.

Case report

An 18-year-old woman born with multiple congenital anomalies and complex arthrogyposis presented with a chief complaint of progressive scoliosis, back pain, postural deformity, and difficulty in breathing on exertion. The patient underwent multiple surgical procedures to both her hands and feet as well her armpits bilaterally. Her karyotype is 46 XX. She was originally diagnosed with scoliosis at age 10 and was treated conservatively. Over the ensuing 8 years, her scoliosis worsened, which ultimately led to impaired respiratory function and functional disability.

On clinical examination, the patient had all the characteristic features of multiple pterygium syndrome. She was of short stature, with flexion deformities of the wrists and hands and webbing across all flexion creases. Bilateral foot deformities were present with a right-sided rockerbottom foot deformity and a residual club foot on the left. Obvious thoracic kyphoscoliosis with significant spinal decompensation was appreciated, with a plumb line of 10 cm from the occiput to the gluteal fold. The deformity was accentuated without a shoe lift. There was more than 30° of rotational prominence at the right thoracic level on forward bending. The patient's left leg was 4 cm shorter than the right (Fig. 1).

There appeared to be good motor strength in all groups to manual muscle testing, but there was significant interference due to the contractures. The patella reflexes were absent, but the Achilles and abdominal reflexes were present. The patient's gait was characterized by short shuffling steps; she was unable to sit in a regular chair because of extension contracture bilaterally involving the hip and knee.

At the initial visit, the patient brought x-rays that were done in 1996. On these films, the upper thoracic kyphosis measured 120°, with an 80° left upper thoracic scoliosis and a 70° right lower thoracic scoliosis. X-rays obtained during the initial visit, in 2000, revealed that the patient's curves had progressed in every plane. At that time, the left upper thoracic curve measured 98° and the right thoracolumbar curve measured 116° (Fig. 2).

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Fig. 1. Anterior and posterior standing photographs of patient taken preoperatively, demonstrating significant spinal decompensation

The preoperative thoracic kyphosis measured 170° and the thoracolumbar lordosis measured 130° (Fig. 3).

Magnetic resonance imaging (MRI) showed extensive lung compression and a markedly reduced space between the sternum and the spine (Figs. 4 and 5).

Fig. 2. Preoperative anteroposterior (AP) standing x-rays showing a 98° left upper thoracic curve and a 116° right thoracolumbar curve

Fig. 3. Preoperative lateral standing x-rays demonstrating a 170° kyphosis and 130° lordosis

Fig. 4. Sagittal and coronal MRI both show extensive lung compression and a markedly reduced space between the sternum and spine

The patient underwent extensive preoperative evaluations by a pediatrician, pulmonologist, and otolaryngologist for a presumed difficult airway. The patient was cleared for surgery. The initial preoperative forced vital capacity (FVC) was 23% predicted. A preoperative echocardiogram showed no structural heart disease and an ejection fraction of 73%.

The patient was taken to the operating room in July of 2000. The anterior approach was done first. A right flank incision was made along the fifth and sixth rib and the chest cavity was entered. Internal thoracoplasties were performed on the ribs of T6-T10. A complete discectomy was carried out from T4-T12. Osteotomies were used to decorticate the end plate, bone graft was inserted in the disc spaces, and an apical resection was done, with fibula allograft used as an inlay strut. Grafton putty was used to supplement the

autograft. A chest tube was placed and the chest closed in a sequential fashion. After the anterior spinal fusion, the patient was turned into the prone position. A longitudinal incision from C7 to L4 was performed and dissected along the spinous process and subperiosteally to expose C7-L3. Distally, pedicle screws at L1-L3 were placed bilaterally, except for L1 on the right side. Because of significant lordosis, the interspinous ligaments were removed and

Fig. 5. Sagittal and coronal MRI both show extensive lung compression and a markedly reduced space between the sternum and spine

Fig. 6. Postoperative AP standing x-rays showing a 48° left upper thoracic curve and a 63° right thoracolumbar curve

Fig. 7. Postoperative lateral standing x-rays demonstrate a 98° kyphosis and a 33° lordosis

Fig. 9. Standing lateral x-rays 3 years after surgery demonstrate a 95° kyphosis and a 55° lordosis

osteotomies were performed at T5 to the T12-L1 level. Rods were attached and sublaminar wires, which had been passed along the concave portion of the spine on both the right and left sides, were tightened, translating the spine posteriorly

Fig. 8. Standing AP x-rays 3 years after surgery show a 65° left upper thoracic curve and a 60° right thoracolumbar curve. The arthrodesis appears to be solid both anteriorly and posteriorly

and correcting the scoliosis and the kyphotic deformity as much as possible. A wake-up test was performed, which the patient responded to, and the wounds were irrigated and closed. The patient suffered a total blood loss of 4 L. Total IV fluids were 4,000 mL lactated ringers, 1,000 mL normal saline, 1,500 mL cell saver blood, 4 U packed red blood cells, 250 mL albumin, 6 U of platelets, and 3 U of fresh-frozen plasma. At the conclusion of the procedure, the patient remained intubated and was taken to the pediatric intensive care unit where she was followed up by the pediatric, pulmonary, ear, nose, and throat, and spine services. The patient's postoperative course was complicated by wound infection requiring irrigation and debridement, a urinary tract infection, and a tracheostomy for persistent atelectasis. The patient eventually recovered from all complications. There were never any focal neurologic deficits.

In the immediate postoperative period, the left thoracic curve measured 48° and the right thoracic curve measured 63° (Fig. 6). The kyphosis measured 98° and the lordosis measured 33° (Fig. 7).

The patient was seen intermittently postoperatively and has done remarkably well. Her last postoperative visit was 3 years after the procedure. The FVC had improved to 60% predicted. In addition, all curves were stable radiographically. X-rays at that time demonstrated a 65° left upper thoracic curve, a 60° right thoracolumbar curve, a 95° kyphosis, and a 55° lordosis (Figs. 8 and 9).

The arthrodesis seems to be solid, both anteriorly and posteriorly. Clinically, the patient stands upright with balance that is greatly improved when compared with her preoperative status; she is extremely satisfied with the outcome of her surgery (Fig. 10). She was able to finish art

Fig. 10. Anterior and posterior standing photographs taken 3 years after surgery demonstrate significant improvement in spinal balance

school and continues her functional activities without difficulty.

Discussion

Some newborns and children with arthrogryposis have webs or pterygia across the extremities; however, there are specific conditions with marked webbing that are seen as a separate entity. Multiple pterygium syndrome (Escobar syndrome) is characterized by flexion contractures at birth. Although webs may not be present at birth, they eventually develop at the neck, elbows, knees, and intracural areas [1]. Cleft palate, deafness, short stature, and scoliosis are frequently present as well. Most cases are inherited in an autosomal recessive pattern, although other types of inheritance have been described. The disease process is progressive, with about 20% developing decreased pulmonary capacity and increasing spinal deformity [1]. There are only a few case reports in the literature describing the surgical treatment of scoliosis in multiple pterygium syndrome [3]. One report does comment on the need for

early surgical intervention to prevent progression of deformity. However, to our knowledge, no authors have reported on the management of multiple pterygium syndrome in the late stage with severe spinal deformity and restrictive lung disease. This case demonstrates the natural history in an isolated patient with multiple pterygium syndrome treated at a late stage. We think that preoperative planning, with a multidisciplinary team and meticulous surgical technique, can result in a balanced spinal correction and restoration of function while arresting eventual pulmonary compromise and early demise.

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