Popliteal pterygium syndrome (PPS) is a very rare autosomal dominant congenital anomaly. The reported incidence is estimated to be 1 in 300,000 live births, with 1:1 male-to-female ratio. Trelat was the first to discuss this syndrome in 1869. The clinical presentation of this condition is diverse, where patients can present with different combinations of clinical manifestations, including musculoskeletal disorders, more specifically popliteal webbing (58%), cleft lip (58%), cleft palate (93%), lower lip pits (46%), fusion of the digits (50%), genitourinary disorders (37%), and nail malformation (33%). In addition, there are other reported manifestations, such as syngnathia, ankyloblepharon, talipes, and digital reduction defects.

The most striking feature of this syndrome is popliteal pterygium contracture, consisting of a connective tissue band starting from the ischial tuberosity and ending at the calcaneus bone. Such a contracture can severely restrict knee extension, leg abduction, and rotation. Lower limb muscles can be abnormally inserted or even absent. Fingers of the upper and lower limbs can also be affected with syndactyly, digit hypoplasia, and brachydactyly being observed. Genitourinary malformation is also reported in both genders. Among girls, labia majora hypoplasia is most commonly reported. On the other hand, unilateral or bilateral cryptorchidism and absent or bifid scrotum is often reported among boys.

The pathogenesis of PPS is poorly understood. Several theories and hypotheses have been proposed in the literature, one of them proposing that it is due to primary microvascular disorders associated with edema, causing a subsequent alteration of the epithelial tissues resulting in secondary adhesions formation, excessive epithelial growth leading to fusion and secondary mesenchymal malinvolve ment, primary collagen defects, and loss of the apoptotic mechanism.

PPS is clinically diagnosed depending on the presence of the above-mentioned range of manifestations. In the absence of a positive family history, some authors have suggested minimal criteria for the diagnosis of popliteal pterygium syndrome, which include at least three of the following characteristics: cleft palate and/or popliteal pterygium, genital anomalies, ectopic salivary ducts in the lower lip, and digit and nail anomalies.

The primary goal of the surgical operation is to restore a good extension angle of the knee, which includes Z-plasty of the skin, nerve and vessel mobilization, and excision of the fibrous band. Generally, children with PPS have a good prognosis. They are expected to have normal development and cognitive function. Reconstructive procedures are available. However, the prognosis for restoring the physical activity depends on the complexity of the case and the success of reconstructive procedure.

**CASE PRESENTATION**

A 30-month-old female child, medically free, was brought by her family to the plastic surgery department in...
Alazhar University Hospital, New Damietta City for a congenital lower limb deformity that was restricting her ability to walk. Upon evaluation through a full history and physical examination, popliteal pterygium contracture of both knees, bilateral equinovarus, ambiguous genitals, and macrodactyly of the little toe of the right foot were noted. Thus, the diagnosis of popliteal pterygium syndrome was established with a negative family history of similar conditions. Both lower limbs were severely affected, with extensive webs running from behind the knees down to the heels, limiting the extension of both knees. Flexion angle of the knee was 85 degrees in the right knee and 95 degrees in the left knee (Fig. 1). We suggested surgery to reconstruct both limbs, yet the patient’s family refused, and insisted we operate on one limb only. Thus, we decided to operate on the most affected one, which was the right leg in this case.

Steps and Tricks

Preoperative marking of a modified jumping man flap was drawn (crab’s flap), as shown in (Fig. 2). Nerve conduction study was performed to document the preoperative status of the patient. In addition, electromyogram was done by placing a small needle in the desired zone, which was the webbed skin of the knee, starting from the contractured band going down to the calf muscle and carefully observing any contractions if the needle stimulates any nearby nerve branches. Once contractions were observed, a marking of the site was done. In this way, we localized the position of the sciatic nerve branches as they might be situated very superficial.

Under general anesthesia, multiple Z-plasties with central rectangular flaps were elevated, as shown in (Fig. 3). Muscle relaxants were not used to observe the muscular response and the nerve stimulation effect intraoperatively. Dissection was done to protect the sciatic nerve branches that were very superficial, and to elongate the whole nerve trunk, we reached up to the nerve under the piriformis muscle. We intentionally did not reach the maximum tension on the neurovascular bundle to avoid injuring any vessels or nerve branches; thus, no nerve grafts were used.

Flaps settled in place and were fixed with 2/0 Vicryl. Immediate postoperative measurements showed an angle that reached 130 degrees in the operated right knee, which measured 85 degrees preoperatively, with a 45 degree immediate improvement. Wounds were dressed appropriately and the knee was put in a back slap. The patient was closely followed up with monthly changed serial back splinting. The patient was tolerating well. Now the angle reached 150 degrees in relaxation position and with improvement of the bilateral equinovarus with substantial improvement in the limb function, as she is now able to walk with some help (Fig. 4). Further improvements are still expected with time, and the patient was advised to keep using night splinting and to follow up with physiotherapy, as we depended on the theory of tissue expansion by postoperative serial splinting.

DISCUSSION

In the etiology of PPS, a hereditary foundation of autosomal dominant inheritance with variable penetration has been suggested. However, our patient has a negative family history of similar conditions. Aside from that, she is a classic case of PPS with popliteal pterygium contracture of both knees, ambiguous genitals, and macrodactyly of the little toe of the right foot. This is in line with previous research results that showed that the sporadic occurrence of this condition is frequently reported. Several theories and hypotheses have been proposed regarding the pathogenesis of PPS, yet it is still poorly understood. We suggest another theory where we believe that the primary disturbance is attributed to the neurovascular insult that mainly leads to a maldevelopment of the nerve during the embryological life, leading to formation of a short nerve, which then hinders the normal growth of the surrounding tissues, including muscles, tendons, and skin folds, causing all the subsequent anatomical malformation. This goes against the previously
The surgical management of the popliteal pterygium contracture can be difficult due to anatomical variations and abnormal pathways of the nerves. This necessitates careful preoperative assessment of the case with good planning, accompanied with precise dissection intraoperatively. As in our case, the sciatic nerve branches were very short, superficial, and running alongside the fibrous band (Fig. 5), which may lead to a decrease in the ability to distinguish them from the fibrous band, increasing the risk of unintended injury to the nerve. Thus, careful dissection with close observation of any nerve stimulation effects.
| Reference                  | No. Cases | Gender | Age     | Severity | Associated Anomalies                                                                 | Family Hx | Management                                                                 | Outcome                                                                 |
|---------------------------|-----------|--------|---------|----------|-------------------------------------------------------------------------------------|-----------|-----------------------------------------------------------------------------|--------------------------------------------------------------------------|
| Qasim and Shaukat         | 3         | F      | 6 y     | N/A      | Cleft lip and palate, lower lip sinus                                                | +ve       | Operated on at the age of 7 and 10 months for the release of the left and right knee flexion contracture, respectively The left leg deformity was severe and needed to be operated on again at the age of 2 years. Developmentally normal, she has learned to walk and run but she cannot put her heels fully plantigrade |                                                                 |
| Kim et al                 | 1         | M      | 7 y     | N/A      | Cleft lip, deformed palate, lower lip sinus                                          | +ve       | Hamstring tenotomy on the ischial tuberosity, tenotomy of the flexor hallucis longus, and Z-lengthening of the Achilles tendon on the ankle followed by Ilizarov external fixator | Full extension was achieved, yet 15 degrees of knee flexion contracture has occurred |
| Ratbi et al               | 1         | F      | 1 mo    | N/A      | Bilateral cleft lip and palate, two pits on the lower lip, oral synechias, achromic spots, bilateral syndactyly of four to five toes, in addition to phimosis | −ve       | N/A                                                                         | N/A                                                                     |
| Posey et al               | 1         | M      | Neonate | Left 90 right 135                      | Ankyloblepharon, left incomplete cleft lip, micro- and retrognathia, synphalangia, hypoplastic nails, asymmetric syndactyly of the second and third toes on the left foot, and cryptorchidism | N/A       | N/A                                                                         | N/A                                                                     |
| Mubungu et al             | 2         | M      | Newborn | N/A      | Hypoplastic nails, asymmetric syndactyly of the second and third toes on the left foot, and cryptorchidism | +ve       | No surgical intervention was done                                          | Lost to follow-up                                                         |
| F                         | 16 y      | N/A    |         |          | Fibrous bands extending from the mid-thigh to the heels, bilaterally, and a pyramidal skinfold covering part of the right hallux's nail | +ve       | N/A                                                                         | N/A                                                                     |
| Katsube et al             | 1         | F      | 3 y     | N/A      | Bilateral cleft lip and palate, lower lip pits, right hand syndactyly, bilateral foot cutaneous syndactyly, bilateral equinovarus, and hypoplastic labia          | +ve       | Bilateral popliteal pterygium repair by Z-plasty and bilateral equinovarus repair when the baby was 11 months old | N/A                                                                     |
| Butali et al              | 2         | N/A    | NA      | Only right knee                        | Complete left unilateral cleft lip and palate                                          | +ve       | N/A                                                                         | N/A                                                                     |
| F                         | N/A       | Only right knee |         |          | Right unilateral cleft of the primary palate                                         | +ve       | N/A                                                                         | N/A                                                                     |

(Continued)
Table 1. (Continued)

| Reference | No. Cases | Gender | Age  | Severity                          | Associated Anomalies                                                                 | Family Hx | Management                                      | Outcome                  |
|-----------|-----------|--------|------|-----------------------------------|--------------------------------------------------------------------------------------|-----------|------------------------------------------------|--------------------------|
| Lees et al. | 9         | F      | 26 y | Ninety degrees bilaterally        | Syndactyly of toes 2-4. Absence of the labia majora and abnormal distribution of pubic hair. Small mouth, submucous cleft palate was present. | +ve       | Managed conservatively with regular physiotherapy | N/A                      |
|           |           | M      | N/A  | N/A                              | Pyramidal folding of tissue over the first toenail                                | +ve       | The web had been corrected surgically            | Resulted in some peroneal nerve damage, with abnormal sensation and sweating in the foot |
|           |           | M      | N/A  | Right knee only and minimal popliteal webbing bilaterally | N/A                                              | +ve       | Treated surgically                                | Having a very active life | N/A
|           |           | F      | N/A  | Oral synchia, unilateral cleft lip and palate, syndactyly of fingers 2/3/4/5 on both hands | N/A                                              | +ve       | Underwent multiple operations over the first 12 years of life | Developmental progress was normal. Although he does have some physical limitations, he now walks well and is fully mobile | N/A
|           |           | F      | 21 y | She was able to extend both knees fully. Linear thickening of the tissues extending from the upper leg to the foot was seen dorsally, more extensive on the left side | Cleft palate and microform cleft lip bilateral lower lip pits. Minimal skin syndactyly of the left hand. The left big toenail was smaller than the right. Double left second toenail. Labia majora were absent. Pubic hair was noted to extend onto the inner upper thighs | –ve       | Surgical release of the tendons was performed at the age of 18 months | N/A                      |
|           |           | N/A    | N/A  | Oral synchia, a unilateral cleft lip and palate with lower lip pits, cryptorchidism. Syndactyly of toes. | N/A                                              | +ve       | N/A                                              | N/A                      |
|           |           | M      | N/A  | Bilateral cleft of the lip and palate, lower lip pits, oral synchia, and ankyloblepharon. Cryptorchidism, syndactyly of toes. | N/A                                              | +ve       | N/A                                              | N/A                      |
|           |           | F      | N/A  | Bilateral cleft of the lip and palate, lower lip pits, hypoplasia of the labia majora, and syndactyly of toes 2/3/4 | N/A                                              | +ve       | N/A                                              | N/A                      |
| Mahalik and Menon19 | 1         | M      | Newborn | N/A                          | Penoscrotal transposition, bifid scrotum with bilateral descended testes, and proximal penile hypospadias were noted. A large mucosal patch was seen in the perineum on the left side. Low posterior hairline, mild ptosis of the right lid, prominent nasal bridge, micrognathia, short neck with ptgym. Dorsolumbar scoliosis, camptodactyly. | N/A       | N/A                                              | N/A                      |
| Penchasza-deh and Salsberg24 | 2         | F      | 17 y | Left 135, operated on before, right did not impend extension | Severe joint contractures of the shoulders, elbows, wrists, fingers, and knees low posterior hairline, low set posteriorly rotated ears, antimongoloid slant of the palpebral fissures, long philtrum, downward turned mouth, and micrognathia. The palate was high and arched, but there was no cleft antecubital pterygia | N/A       | N/A                                              | N/A                      |
|           |           | M      | 6 y  | Previously operated on            | N/A                                              | N/A       | N/A                                              | N/A                      |

(Continued)
When planning the procedure, we decided to do a modified jumping man flap, which we called the crab’s flap, with a central rectangular flap to completely cover the popliteal fossa and to prevent contractions (Fig. 2). As a result, we had enough skin to cover the surgical site without facing any problems.

The improvement in our patient was not achieved only by surgical intervention, to avoid tension over the nerves that might cause nerve injury. Thus, serial splinting was done monthly postoperatively, as we believed in tissue expansion by serial splinting following surgical intervention. The extension angle of the right lower limb had increased from 85 degrees to 130 degrees with 45 degrees improvement; such improvement was considered good without injuring the nerve, as this was the limit. Of course, various factors play a role in the extent of the surgical outcome with the length of the sciatic nerve in the free edge being the limiting factor in this procedure. Every procedure should be personally tailored to fit each patient. Serial back splinting and physiotherapy had a substantial role in the postoperative improvement in our case, where the angle has improved an additional 20 degrees, reaching 150 degrees in total. Further improvements are expected with time. The functional aspect of the patient was the most important to us, as the patient’s ability to walk was restored. We still expect to see more improvements with time, as there is still extension lag about 30 degrees, and the patient was advised to keep using night splinting and to follow up with physiotherapy.

In addition, a comprehensive review of the literature, using the PubMed search engine of the National Library of Medicine and National Institutes of Health, regarding popliteal pterygium syndrome presenting with knee webbing has been conducted to discuss various factors of this condition and to provide an updated summary of all similar reported cases. The following terms were searched for: (popliteal pterygium syndrome) OR (popliteal pterygium contracture). Among 255 articles, only 13 were included, which reported a total of 27 cases of PPS with knee webbing. Some factors were not mentioned in the published reports; thus, we indicated such missing information with “not applicable” (N/A). The male-to-female ratio was 12:13, with two cases not reporting the gender. In total, 17 cases were familial, only two cases were sporadic, and the remaining did not specify. Postoperative serial splinting was not performed in any of the cases. Yet, some other methods were occasionally reported. Hyoung used an Ilizarov external fixator, which reached full angle of extension postoperatively, yet they reported that 15 degrees of knee flexion contracture occurred later on.15 In addition, Surya used plaster of Paris slab in the postoperative period for 1 week with his patient.16 All included cases are listed and summarized in Table 1.

CONCLUSIONS

Popliteal pterygium syndrome is a very rare condition that involves several malformations, with hereditary basis...
being suggested. However, sporadic cases such as in our patient are usually seen.

The procedure should be carefully planned, and the exact position of the sciatic nerve branches should be identified, as they might be situated very superficial. Thus, nerve conduction studies and EMG should be done preoperatively to assess the exact position of the nerve and muscle function. Combining a central rectangular flap with Z-plasties can be beneficial to prevent contraction and to completely cover the popliteal fossa. In addition, we recommend postoperative serial splinting to be considered in the management plan, as it showed significant additional improvement in the extension angle.

Mohammed Sewilam, MD
Jubail industrial City
Saudi Arabia
E-mail: mohammedplast48@gmail.com

REFERENCES
1. Melnick M, Bixler D, Fogh-Andersen P, et al. Cleft lip+/−cleft palate: an overview of the literature and an analysis of Danish cases born between 1941 and 1968. Am J Med Genet. 1980;6:83–97.
2. Froster-Iskenius UG. Popliteal pterygium syndrome. J Med Genet. 1990;27:320–326.
3. Bixler D, Poland C, Nance WE. Phenotypic variation in the popliteal pterygium syndrome. Clin Genet. 1973;4:220–228.
4. Spencer LSB, Gondim DD, Alves RV, et al. Popliteal pterygium syndrome: case report and literature review. Rev Bras Cir Plást. 2012;27:482–486.
5. Lees MM, Winter RM, Malcolm S, et al. Popliteal pterygium syndrome: a clinical study of three families and report of linkage to the Van der Woude syndrome locus on 1q32. J Med Genet. 1999;36:888–892.
6. Herold HZ, Shmuoli G, Baruchin AM. Popliteal pterygium syndrome. Clin Orthop Relat Res. 1986;209:194–197.
7. Orpha.net. Autosomal dominant popliteal pterygium syndrome [online]. Available at: https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=1300. Published 2019. Accessed 30 March 2021.
8. Haramis HT, Apesos J. Cleft palate and congenital lateral alveolar synchia syndrome: case presentation and literature review. Ann Plast Surg. 1995;34:424–430.
9. Spauwen PH, Cobben JM, Gardeniers JW. The popliteal pterygium syndrome: an analysis of two families. J Craniomaxillofac Surg. 1994;22:276–280.
10. Koch H, Grzonka M, Koch J. Popliteal pterygium syndrome with special consideration of the cleft malformation: case report. Cleft Palate Craniofac J. 1992;29:80–84.
11. Khan SN, Hufnagle KG, Pool R. Intrafamilial variability of popliteal pterygium syndrome: a family description. Cleft Palate J. 1986;23:233–236.
12. Gahm C, Kuylenstierna R, Papatziamos G. Popliteal pterygium syndrome (PPS) with intra-alveolar synchytia: a discussion of anesthetic and surgical considerations. Int J Pediatr Otorhinolaryngol. 2007;71:1613–1616.
13. Escobar V, Weaver D. Popliteal pterygium syndrome: a phenotypic and genetic analysis. J Med Genet. 1978;15:35–42.
14. Ferreras JM, Pimentel CR, Vasquez ZA, et al. Syndrome de pterigion popliteo: a proposito de un caso. Acta Médica Dominicana. 1992;14:20–22.
15. Kim HM, Park IJ, Jeong C. Treatment of popliteal pterygium using an Ilizarov external fixator. Clin Orthop Surg. 2009;1:236–239.
16. Venkata Mahipathy SRR, Durairaj AR, Sundaramurthy N, et al. Popliteal pterygium syndrome: a rare syndrome. Indian J Plast Surg. 2018;51:248–250.
17. Qasim M, Shaukat M. Popliteal pterygium syndrome: a rare entity. APSP J Case Rep. 2012;3:5.
18. Ratbi I, Fejil N, Legendre M, et al. Clinical and molecular findings in a Moroccan patient with popliteal pterygium syndrome: a case report. J Med Case Rep. 2014;8:471.
19. Posey JE, Dariya V, Edmonds JL, et al. Syngnathia and obstructive apnea in a case of popliteal pterygium syndrome. Eur J Pediatr. 2014;173:1741–1744.
20. Mubungu G, Lumaka A, Matondo R, et al. Skinfold over toenail is pathognomonic for the popliteal pterygium syndrome in a Congolese family with large intrafamilial variability. Clin Case Rep. 2014;2:250–253.
21. Katsube M, Yoshiura K, Kusumoto K. A Japanese family with popliteal pterygium syndrome. Case Reports Plast Surg Hand Surg. 2015;2:50–52.
22. Batali A, Mossey PA, Adeyemo WL, et al. Novel IRF6 mutations in families with Van Der Woude syndrome and popliteal pterygium syndrome from sub-Saharan Africa. Mol Genet Genomic Med. 2014;2:254–260.
23. Mahalik SK, Menon P. Popliteal pterygium syndrome. J Indian Assoc Plast Surg. 2010;15:110–111.
24. Penchazadeh VB, Salszberg B. Multiple pterygium syndrome. J Med Genet. 1981;18:451–455.
25. Sethi P, Bhatia PK, Gupta N, et al. Multiple pterygium syndrome: challenge for anesthesiologist. Saudi J Anaesth. 2016;10:350–352.