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A case of popliteal pterygium syndrome

Popliteal pterygium sendromu: Olgu sunumu

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Popliteal pterygium sendromu nadir görülen, genellikle otozomal dominant geçiş gösteren bir hastalıktır. Dört yaşında getirilen bir kız çocuğunda sol taraftaki popliteal bölgedeki perdeli deri uzantısı gluteal bölgeden topuğa kadar uzanmaktaydı ve ilave olarak inkomplet yarık dudak, hipotrofik labialar ile ayak/parmak/tırnak anomalileri vardı. Yapılan "Z " plasti girişimleriyle dizdeki deformite düzeltildi. Ancak daha sonra hasta ile olan bağlantı kesildi. On yıl sonra hasta tekrar getirildiğinde sol tibiasında yaklaşık 15 cm'lik kısalık ile ayak deformitesinin nüks ettiği görüldü. Bunun üzerine dizindeki kontraktür düzeltilerek hastanın da istemesiyle diz altı amputasyon uygulandı. Hastaya protez planlandı. Ancak hasta ile olan ilişki tekrar kesildi ve hasta ve yakınlarına ulaşılamadı.

Anahtar sözcükler: Yarık damak; kontraktür/konjenital/cerrahi; ekstremite/anormallik; diz/anormallik; pterygium/cerrahi.

Popliteal pterygium syndrome is a rare disorder, usually showing autosomal dominant inheritance. A four-year-old girl presented with a popliteal web on the left extending from the buttock to the heel. She also had an incomplete cleft lip, hypoplastic labia majora and foot/digit/nail anomalies. The popliteal web was released by Z-plasty procedures. However, the patient dropped out of further treatment. Ten years later she was brought again with severe deformity in her left lower extremity, tibial shortening of approximately 15 cm and foot deformity. Nearly-full knee extension was obtained and below-knee amputation was performed at the patient's request. Plans of inserting a prosthesis was prevented by the loss of contact with the patient and her family.

Key words: Cleft lip; contracture/congenital/surgery; extremities/abnormalities; knee/abnormalities; pterygium/surgery.

The popliteal pterygium syndrome (PPS) is a rare, usually autosomal dominant disorder. The cardinal features include genitourinary, craniofacial, and extremity abnormalities in association with popliteal web.[1-9] Popliteal web (pterygia) can be in various degrees; it can be a trace, hardly discernible, pigmented line extending posteriorly from the upper thigh to the heel[7] as well as from the ischium to the os calcis (heel bone, calcaneum) sometimes,[1,5,7-9] or even from the gluteal region to the heel.[7] It's commonly bilateral.[1,2,5,6,8,9] Popliteal webbing may cause fixated flexion deformities in knees or club foot deformities in feet.[9] Various degrees of anomalies may be present in other members of the family.[4,7,9] Associated orthopaedic findings include: adactyly, syndactyly, brachydactyly, clinodactyly, dysplastic nails, hypoplastic or absent first rays, contracted heel cords, hypoplastic patellae, scoliosis, rib anomalies, and hypoplastic tibiae.[5,7,9] Additionally club foot deformities, equinus deformities and vertical talus may be present in feet;[9] flexion-adduction deformities may be present in hip,[5,9] spina bifida occulta or lordosis may be present.[5] Despite the present of hamstrings and sural (calf) muscle groups,

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their absence is also reported.[5,7] One should be kept in mind that sciatic nerve is in the subcutenous posterior part of the fibrotic popliteal web. Because of this property, correction of knee contractures can be hard and risky.[5,7,8] Although it is reported that popliteal vessels are present in deeper parts of popliteal region, which is their normal anatomic location, it is also reported that tibial nerve can lie adjacent to posterior margin of fibrotic web (pterygeal fold) with popliteal and posterior vascular structures, in some cases.[9]

Case report

A four-year-old girl was presented to Orthopaedics and Traumatology Department of Dicle University Medical Faculty in 1985, because of her congenital deformity in lower extremity on the left. A skin web was observed including the popliteal region in her left lower extremity, extending from the gluteal region to the heel, which was fixating the knee in full flexion, in clinical examination; it was also observed that the foot was fixated in equinovarus position; finger and nail abnormalities were associating with them (Figure 1). As additional findings, incomplete cleft lip of the lower lip and hypoplastic labia majora, were inspected (440-1575/043653). The parents are first-degree relatives and the other family members do not have a similar congenital deformity, as we have learned from her history. After completing required examinations, contracture of the knee was corrected by extracting the fibrotic band and performing two stages Z-plasty procedures, and by free-skin graft, which was taken from the opposite thigh region (Figure 2). Sciatic nerve and other neurovascular structures did not cause any problem during correction. Correction of the deformity in foot was left to the other stage of the surgery and the patient was discharged from the hospital. Later on, neither the patient was taken to hospital back for the controls, nor answered the control call-ups (the address was booked to her file). The patient referred to our clinic 10 years later, in 1995, because of the repetitive deformity in her left lower extremity. In clinical examination, 20˚ flexion contracture of the left knee and diffuse fibrotic tissue in popliteal region were observed. It was also observed that tibiotalar articular integrity was lost so talus was displaced posteriorly with the foot, distal tibia and the region corresponding to the inferior

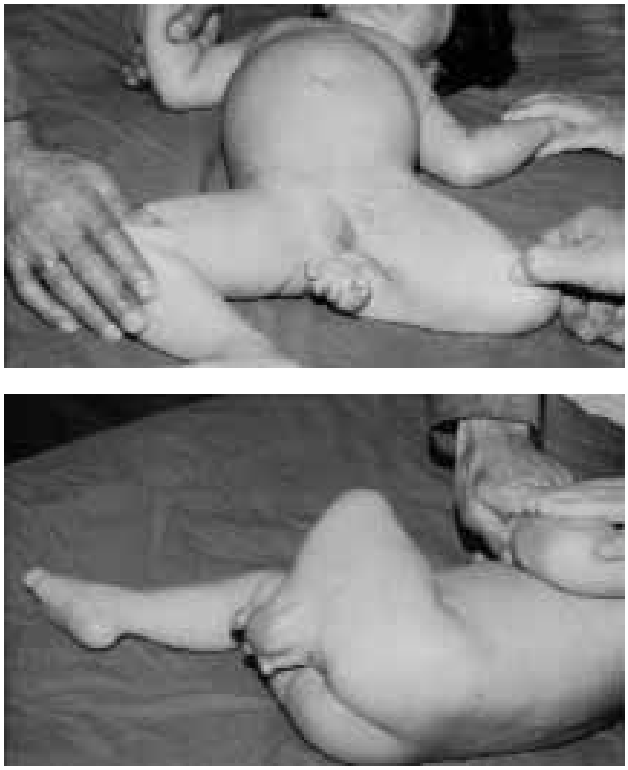
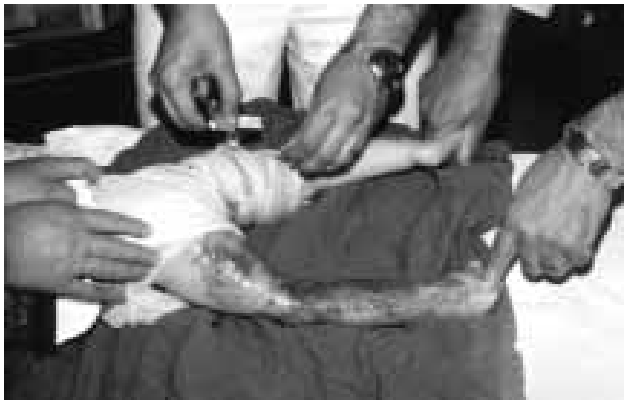


Figure 1. The popliteal web in left lower extremity of the case, fixated knee and foot deformities, finger and nail abnormalities, hypoplastic labiums.

articular surface was compressing the skin and a contracture was developed in plantar region, which was touching the posteromedial part of the crus. 15 cm. of shortness was observed in tibia (Figure 3). When asked “why she was not taken/came to clinic for the controls”, the given answer was not satisfactory. After correction the contracture in her knee, below knee amputation was performed by taking the patients and her relatives consent and approval. A prosthesis application was planned. However we have lost contact with the patient again. We could not get in touch with the patient or her relatives.

Discussion

The popliteal pterygium syndrome is a rare, usually an autosomal dominant disorder.[1-9] Similar findings may exist in other family members or siblings, especially.[1-4,9] The disease has two forms. It is reported that the mild form of the disease is inherited autosomal dominantly, severe form is inherited autosomal recessively and additional multiple anomalies may be present.[6] Deskin and Sawyer[2] stated that, most of the cases are sporadic and in less than



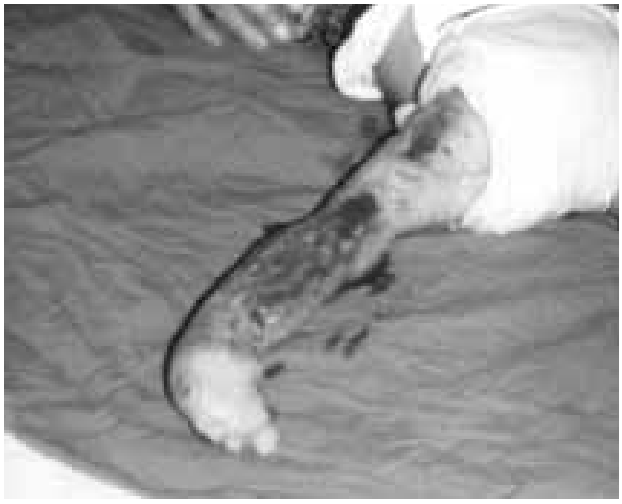
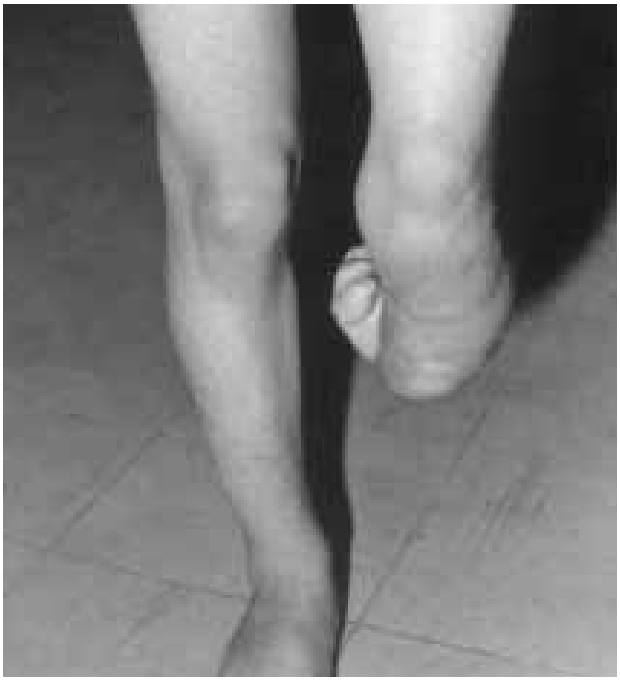


Figure 2. Early postoperative views of the same case, after two stage-Z plasty and correcting the contracture by taking free skin graft from the order thigh.

1/3 of the cases, a few family members are affected. Hamamoto and Matsumoto,[6] in a case which they have presented, reported that the other members of the family doesn't have any congenital abnormality, siblings are normal and, mother and father are not relatives. In our case, patients' mother and father stated that, other members of the family do not have any similar congenital abnormality. Mother and father were first degree relatives.

The popliteal web, which is a fibrotic tissue, extends from the ischium to the calcaneus; sometimes closes up the heel to the gluteal region (buttock). The foot may be in fixed equinus position and the knee may be in fixed flexion contracture position.[7] A fully fixed flexion knee contracture was present in our case too and heel was in contact with ischium/gluteal region (buttock). Popliteal web was very close to anal region. Additionally, there were



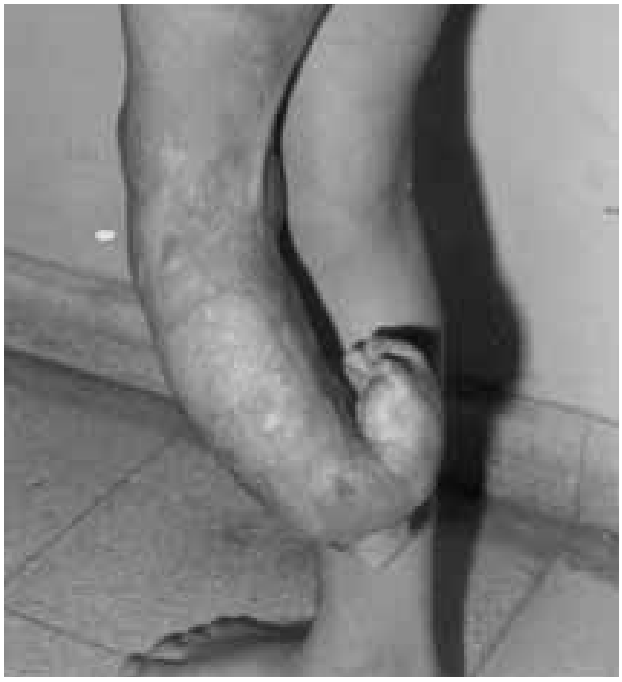


Figure 3. Left knee, crus and foot deformity views of the same case after 10 years, who did not refer to clinic for the follows up.

fixed equinus and varus deformities in foot (Figure 1).

The sciatic nerve may be shortened and may extend close to fibrotic band more posteriorly, which is an unusual location for it. The shortened sciatic nerve may limit the surgery, providing extension.[5,7,8] In our case, two stages Z-plasty procedures were performed after extracting the fibrotic band and sciatic nerve did not cause any handicap during correction of the contracture.

It's reported that only popliteal web existence, is not enough for the diagnosis. Popliteal web may be an associated finding in abnormalities like arthrogryposis,[2,7,10] sacral agenesis,[10] lethal popliteal pterygium syndrome,[2] popliteal pterygium associated with ectodermal dysplasia[2] and multiple pterygia.[2,3] In our case, incomplete lower cleft lip, hypoplasic labia majora, equinovarus deformity in foot and finger/nail anomalies were present and we evaluated this case as popliteal pterygium syndrome.

Various methods are applied and described in treatment. Hanson (1914), wanted to apply surgery in a popliteal pterygium syndrome patient, however, he abandoned the early surgical application because of the abnormal location of sciatic nerve and treated the patient with casts and splints for several years; where as Hackenbrock (1924) performed a 6-cm shortening femoral osteotomy to his popliteal pterygium syndrome patient who is having 90 degrees of flexion contracture.[7] 70 degrees of flexion contracture was remained in one of the two cases who applied Z-plasty with extracting fibrotic band and physiotherapy after serial cast corrections in postoperative period, by Champion and Cregan.[1] Oppenheim et al.[7] reported that lengthening the Achilles tendon would be efficient in a case where popliteal web causes a mild deformity; where as Z-plasty is performed to release the soft tissue in moderate or severe deformities, shortening femoral osteotomy is additionally performed to correct the flexion contracture; however, in different periods they applied knee disarticulation to one side and below-knee amputation to the other side of another case having bilateral popliteal pterygium syndrome and multiple anomalies. The shortening osteotomy that would be performed to 1/3 distal of the femur is also recommended by the other authors.[8,10] Only soft tissue procedures alone, can give good results in cases where the sciatic nerve is sufficient length to permit full or near full knee extension.[7] Besides these, there are authors, who report that amputation might be necessary in cases where the flexion degree is severe, in contemplation of popliteal vessels and nerves may be shortened.[10] The extremity shortness, developing in unilateral cases, may cause problems in forthcoming periods.[9] We have corrected the flexion contracture in our case by two stages Z-plasty. No postoperative neurovascular problem developed. But the patient was out of our follow ups. If there weren't any follow up problems, with the early diagnose of the foot deformity and shortness in her last control, it might be possible to prevent or treat the deformity with external circular fixator or with a similar method and compensate the shortness.

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