# Popliteal Pterygium Associated with Atrial Septal Defect. A Case Report

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## Intruduction

Popliteal pterygia are found in popliteal pterygium syndrome (PPS), multiple pterygium syndrome, arthrogryposis, and other rare conditions [1]. Diagnosis of PPS is based on a classical triad (craniofacial, genitourinary, and extremity anomalies, including the popliteal pterygium) and genetic malformations [2-3]. Cleft palate, with or without cleft lip, has been found to be the most frequent feature in PPS [1-5].

Here we report a case of typical popliteal pterygium, underdeveloped vagina, talipes planovalgus, hyperpigmentation, hypoplastic teeth, and atrial septal defect (ASD). No cleft palate or cleft lip was present. This combination of anomalies has not been reported previously.

## Case Report

A girl (46,XX) was born as the second child of healthy parents (a 34-year-old father and 30-year-old mother) in week 38 of gestation. Her eight-year-old brother showed no congenital anomalies. The delivery was unremarkable, with a birth weight of 2,690 g and an Apgar score of 9 points. Examination showed popliteal webbing, underdeveloped vagina, hypertrophy of the clitoris, talipes planovalgus, and a hyperpigmented skin lesion in the lumbar area. Posterior webbing on the right leg extended over the popliteal region from the lateral part of the forefoot to the ischial tuberosity, contracting the knee joint. Passive range of motion was -90° extension and 130° flexion (Fig. 1A, B). Talipes planovalgus was noted in the right foot but could be corrected by manipulation in the knee-flexed position. Nail anomalies or syndactyly of the hand or feet were not observed.

Findings by simple X-ray imaging were unremarkable, and no abnormal findings such as hypoplasia or joint fusion were observed. A systolic murmur was audible around the 2$^{nd}$ LSB, and ultrasound cardiography revealed a small ASD (4.8mm diameter) and LR shunt. No RV dilatation or VSD was detected. Right popliteal ultrasound showed no major blood vessels at the apex of the pterygium. Instead, the vessels were localized within the pterygium, somewhat further back than normal. Sequence genetic analysis showed no anomalies in interferon regulatory factor 6 (IRF6) on 1q32 [2-3].

Z-plasty procedures were performed to release the popliteal web when the child was 6 months of age. The apex of the pterygium contained only collagen fibers, with the shortened nerves and blood vessels located deeper within the pterygium. The knee joint was extended carefully to avoid separating or placing excessive stress on the neural and vascular tissue. The child's subsequent growth and development progressed within the normal range, although the knee retained a flexion contracture. Between 2 and 3 years of age, she underwent a dermatoplasty and surgical lengthening of the Achilles tendon. However, tight neurovascular bundles obstructed the full extension of the knee joint. The ASD had reduced spontaneously, although a mild systolic murmur remained. The child is now 8 years of age. The knee joint has -20° of extension (-10° for passive movement) and 140° of flexion, and the ankle joint has -20° of dorsi-flexion and 30° of plantar-flexion. Her right leg is about 4 cm shorter than her left, but muscle strength is adequate and she is able to attend elementary school. Imaging findings show no signs of osteonecrosis or of osteoarthritic changes, and activities of daily living are unimpaired (Fig. 2). Her upper and lower teeth are smaller than normal and deformed, but her chewing ability is not markedly impaired.

## Discussion

Popliteal pterygia are found in popliteal pterygium syndrome (PPS), multiple pterygium syndrome, arthrogryposis, and other rare conditions [1]. Popliteal pterygium syndrome is a rare autosomal dominant disorder in a 1:1 male: female ratio, with an incidence of 1/300,000 [3-6]. The diagnostic criteria demand at least three of five abnormalities: (1) cleft lip or palate, (2) popliteal pterygium, (3) paramedian lower lip sinuses, (4) genitourinary anomalies, and (5) extremity anomalies [1-5]. Our case had popliteal pterygia, underdeveloped vagina, hypertrophy of the clitoris, talipes planovalgus, hyperpigmentation, and ASD. She was free, however, from cleft palate or cleft lip or exonic mutation in IRF6.

There are challenges in reaching a critical diagnosis of PPS based solely on clinical and molecular features. In a

review of the literature presented by Oppenheim et al. [4], only 6 (9%) of PPS patients were free of craniofacial abnormalities.

The complication of cardiac defect in this patient is also seen in cases of multiple pterygium syndrome (MPS) [6-8]. This case did not satisfy the diagnostic criteria for MPS, however, as distinct facial features and respiratory disorders were absent, and as the pterygium and joint contracture were monostotic. Similarly, the features and etiology of our case could not be associated with arthrogryposis or with other rare conditions such as Noonan syndrome, prune belly syndrome, Klippel-Feil syndrome, LEOPARD syndrome, or fetal akinesia syndrome. With all these etiological possibilities, we are left with no clear etiological factors. Ultimately, the patient was diagnosed with an extremely rare case of popliteal pterygium combined with ASD.

The therapeutic strategy for popliteal pterygium is difficult, and multiple reconstructive surgeries are required [4,9-12]. In particular, considerable knee extension disability may remain after reconstructive surgery in cases where the shortened sciatic nerve and popliteal artery are involved at the apex of pterygium. In this case, the shortened neurovascular bundles were deep within the pterygium. This made it possible to improve the range of motion of the knee to -20° extension by applying Z-plasty procedures to soft tissue such as the skin, fascia and ligaments, and performing Achilles tendon-lengthening surgery. There are no apparent indications for follow-up surgery at present. Further intervention may be necessary, however, in the future.

## References


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## Explanation of Figures

Fig. 1 shows the initial findings for the right leg.
A: full extension
B: flexion
Extension of the right knee joint was restricted by the posterior pterygium.
Fig. 2 Appearance at 8 years of age
There are visible restrictions on knee joint extension and on dorsi-flexion of the ankle joint.

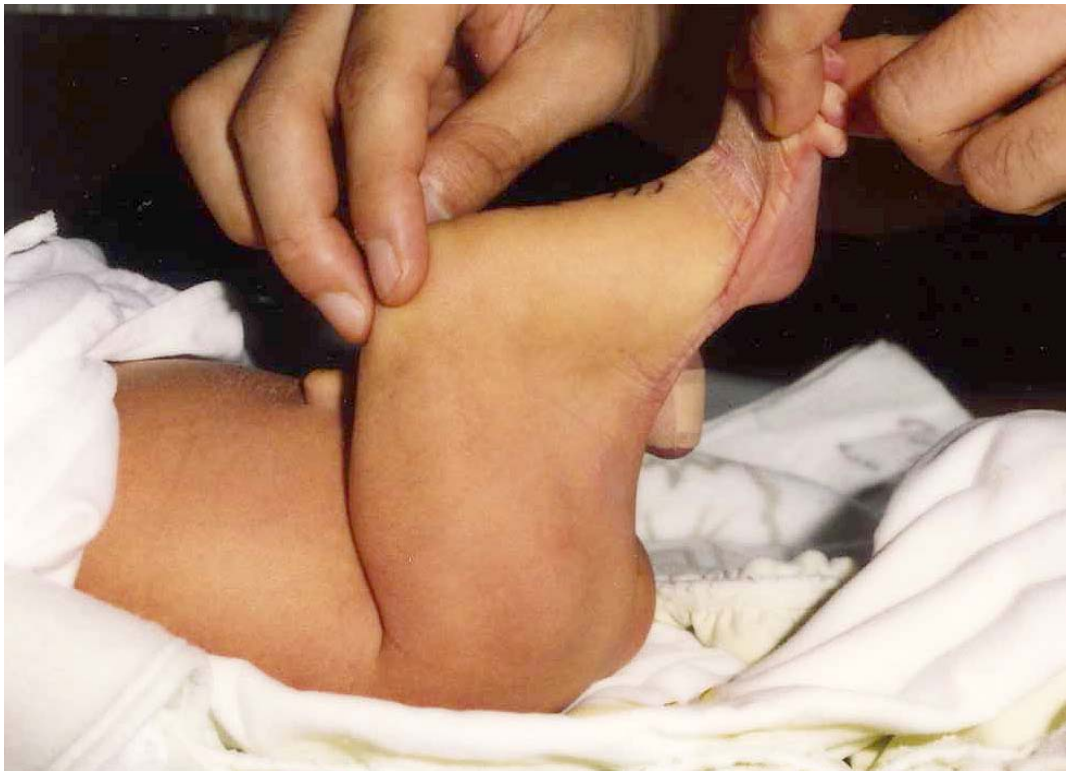

Figure 1A

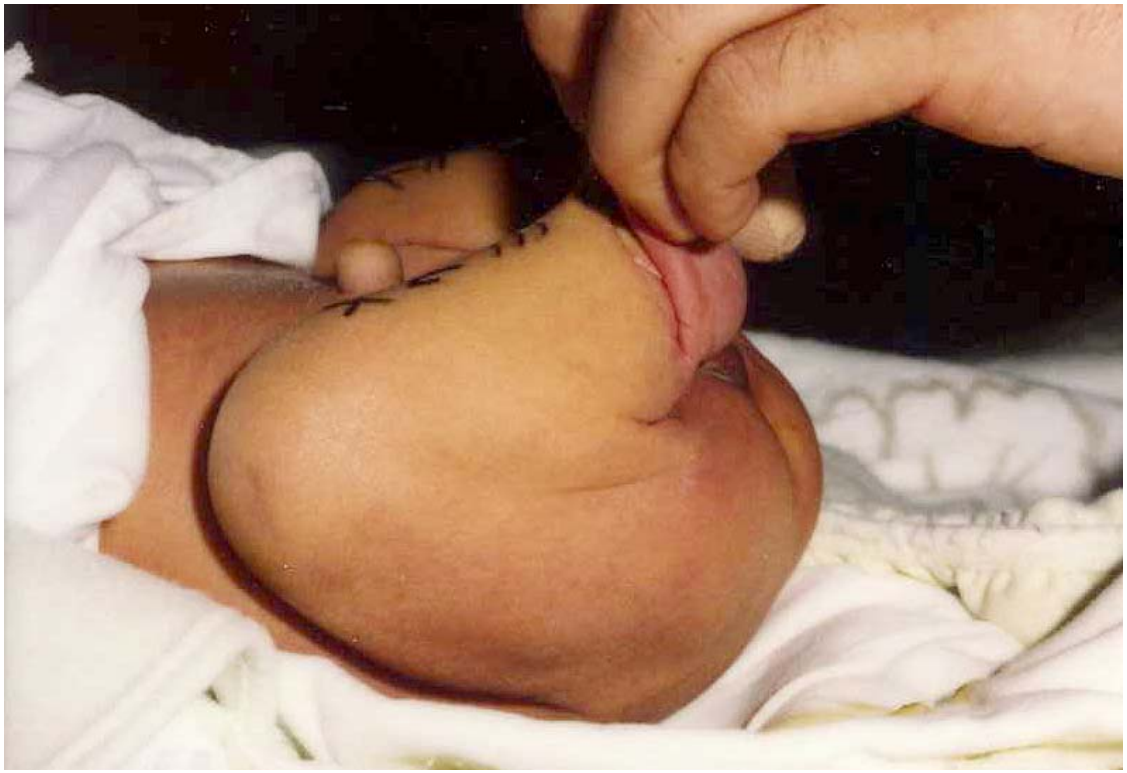

Figure 1B

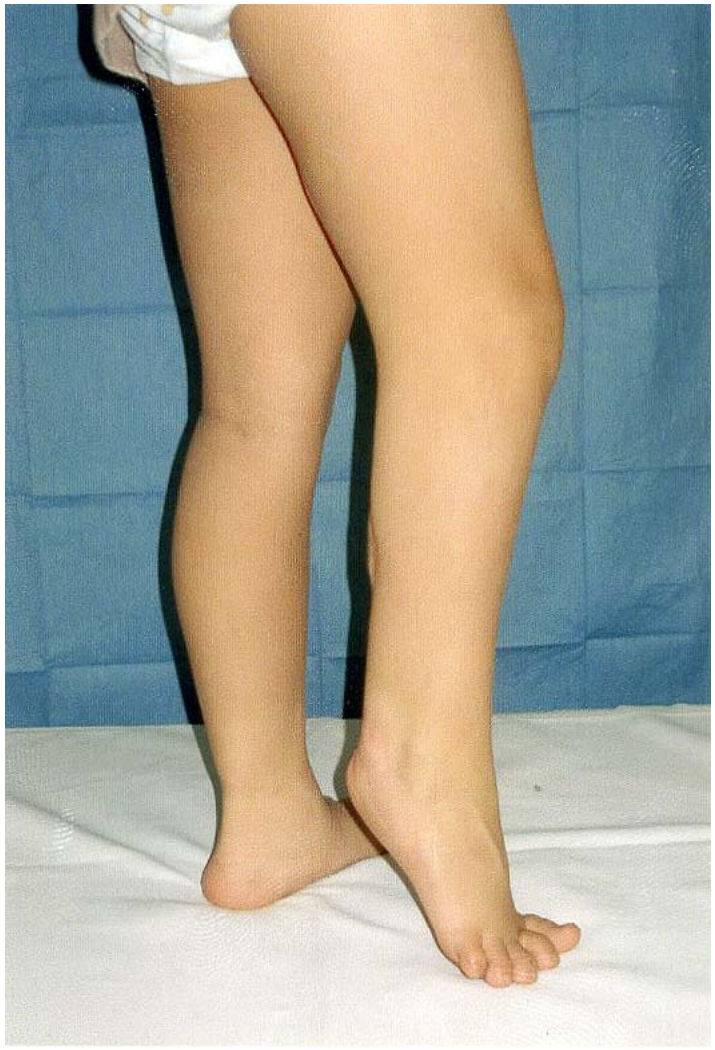

Figure 2