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CASE REPORT
Craniofacial/Pediatric

Popliteal Pterygium Syndrome: A Case Report Highlighting Challenges and Surgical Interventions in a Resource-limited Setting

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Summary: Popliteal pterygium syndrome is a rare congenital disorder characterized by facial, genitourinary, and musculoskeletal anomalies, with popliteal webbing being notably challenging. A 4-year-old boy presented with progressive limping, cleft palate, and genital malformations. He had no follow-up care after an intraoral band excision at 15 days old. The boy underwent surgery for left-sided popliteal webbing, followed by genital and cleft palate repair. A modified jumping man Z-plasty flap was used for the popliteal webbing, followed by splinting. Subsequent follow-ups showed no complications. Enhancing care in resource-constrained settings requires addressing challenges such as delayed interventions due to late follow-up, limited awareness between communities and healthcare professionals, social stigma, and inadequate healthcare understanding. These obstacles hinder timely diagnosis and intervention, underscoring the need for increased awareness and effective early intervention strategies. Early detection and parental counseling are critical in managing popliteal pterygium syndrome. Timely surgical planning, including addressing orofacial and genital deformities and using Z-plasty for webbing release, is essential. Postoperative splinting significantly improves outcomes.
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Popliteal pterygium syndrome (PPS) is an exceedingly rare congenital anomaly, inherited in an autosomal dominant manner, affecting approximately 1 in 300,000 live births. Initially described by Trelat in 1869, the condition was later named PPS by Gorlin et al due to its distinctive feature, which is popliteal webbing.[1,2]

PPS encompasses a spectrum of orofacial, cutaneous, musculoskeletal, and genitourinary anomalies, presenting highly variable clinical features. Common anomalies include cleft palate (93%), cleft lip (58%), lower lip pits (46%), popliteal webbing (58%), syndactyly (50%), and genitourinary anomalies (37%). Despite these deformities, growth and intelligence typically remain unaffected. PPS has been reported in both sporadic and familial cases, with familial cases often displaying autosomal dominant transmission, decreased penetrance, and significant intrafamilial phenotypic variation.[2]

PPS shares clinical features with Van der Woude syndrome, with PPS being the more severe form.[3,4] Mutations in the *IRF6* gene have been identified as the causative factor for both Van der Woude syndrome and PPS. The differential diagnosis includes syndromes with orofacial anomalies and limb defects.[5] Although PPS is rare, its complexities necessitate comprehensive case documentation to enhance understanding and improve management strategies.[2,6,7]

CASE PRESENTATION

A 4-year-old boy presented with progressive limping. The child was born via spontaneous vaginal delivery, with no notable issues reported during the pregnancy. At 15 days old, he underwent surgery to remove an intraoral band but had no follow-up care. As he grew, his family noticed atypical features on his lower extremities, genitalia, and palate but did not seek further medical evaluation until his limp worsened.

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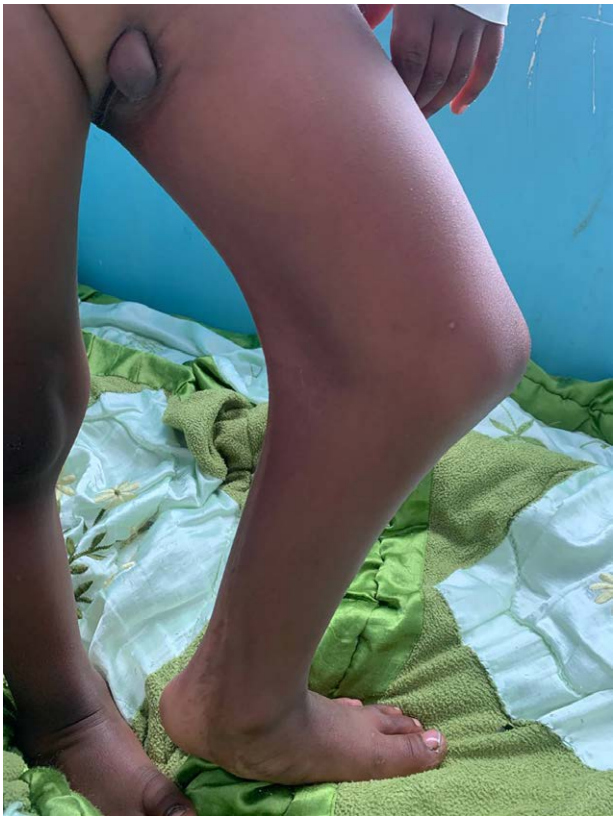

Fig. 1. Preoperative photograph of the patient showing left popliteal pterygium.

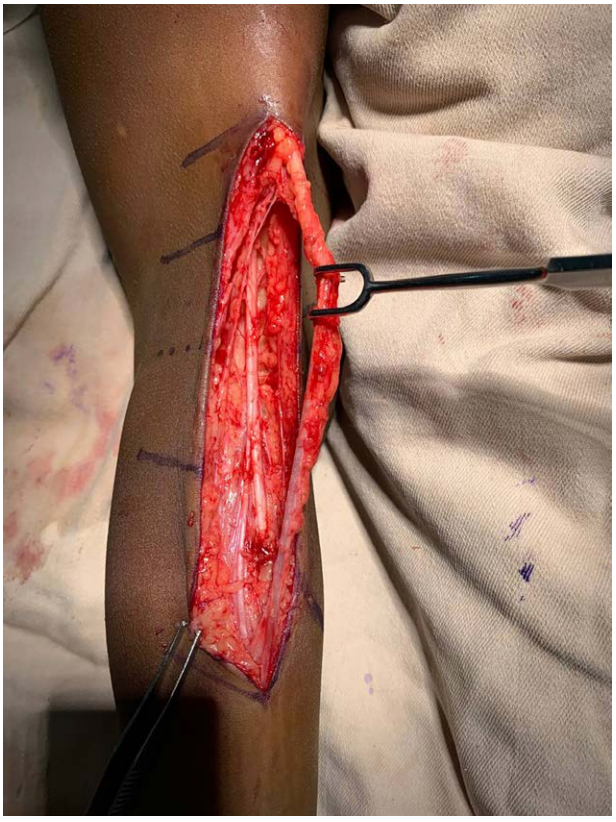

Fig. 2. Intraoperative photograph showing the contracting band (held by hook) and underlying popliteal nerve.

Upon examination, the boy exhibited lower lip pits, a cleft palate, genital anomalies, and significant popliteal webbing, particularly on the left side. His growth and neuropsychomotor development were normal, but his speech was impaired due to the cleft palate. Physical examination revealed bilateral moist lower lip pits, a Veau type-II cleft palate, and a flexion contracture of the left knee, preventing the child from placing his left foot flat on the ground (Fig. 1).

The patient's family opted for the left-sided popliteal webbing release as the initial procedure. Preoperative preparations were completed, and a modified jumping man Z-plasty flap (crab flap) was marked and executed. During surgery, a contracting subcutaneous cord extending from the proximal thigh to the posterior heel was observed (Fig. 2). The sciatic nerve, shortened and adhered to the cord, was carefully dissected, released, and protected. The limb was nearly fully extended after the nerve mobilization, and the flaps were precisely inset (Fig. 3). The limb was immobilized with a posterior gutter splint, and splinting was planned for 3 weeks.

At follow-up, the patient showed significant improvement, walking in an upright position (Fig. 4). Satisfied with the outcome, the family consented to further procedures, including cleft palate repair and genital reconstruction, which were successfully performed. The family is now engaged in regular follow-up visits, with overall positive outcomes.

DISCUSSION

PPS is a hereditary condition with autosomal dominant inheritance and variable penetrance.[8] This case presented the classic features of PPS, including popliteal pterygium, cleft palate, lower lip pits, genital anomalies, and foot deformities. The absence of a family history aligns with the sporadic form of the syndrome, which is most commonly reported. Despite lacking timely follow-up, the patient's growth and development were unaffected, consistent with previous studies.[6,7,9]

The lack of early follow-up led to missed interventions, such as palatoplasty and popliteal web release, which could have been performed earlier. The case underscores the importance of early parental counseling and the challenges faced in resource-limited settings, where cultural beliefs, social stigma, and inadequate healthcare contribute to delays in treatment.[6,9]

Digital technologies for monitoring wound healing, such as digitalized measurement systems, could improve care for patients with PPS by enabling cost-effective, flexible, and reproducible monitoring of wounds. In addition, addressing factors such as lack of awareness, financial constraints, and limited access to specialized care are crucial for the timely and optimal treatment of orofacial clefts and other PPS-related anomalies.[10]

In this case, the successful popliteal webbing release encouraged the family to proceed with further surgical interventions. This highlights the importance of

Fig. 3. Immediate postoperative photograph after wound closure using modified jumping man Z-plasty flap (crab flap) design.

Fig. 4. Postoperative photograph taken at 3-month follow-up.

comprehensive counseling and screening for subclinical findings in patients with PPS and their families. Advanced surgical techniques, such as tendon lengthening or the Ilizarov method, may be necessary in severe cases to improve joint mobility.

CONCLUSIONS

Raising awareness about PPS is paramount for effective management. Early detection and parental counseling are essential for ensuring timely interventions. Proper surgical planning, including addressing orofacial and genital deformities and performing Z-plasty for webbing release, significantly improves outcomes. Postoperative splinting plays a crucial role in enhancing limb extension, and a combination of early intervention and increased awareness optimizes care for patients with PPS.

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DISCLOSURE

The authors have no financial interest to declare in relation to the content of this article.

PATIENT CONSENT

Parents or guardians provided written consent for the use of the patients' image.

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