



Popliteal Pterygium Syndrome at the Mother and Child University Hospital Lagune of Cotonou: Epidemiological, Clinical and Therapeutic Aspects

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ABSTRACT

Introduction: Congenital webbing knee or popliteal pterygium is the tissue filling the posterior aspect of the joint limiting extension. It can be part of several syndromes including the popliteal pterygium syndrome (SPP).

Purpose: To present the cases of patients suffering from

Patient and Method: This is a description of epidemiologica, clinical and therapeutic aspects of clinical cases of webbing admitted to our unit over 10 years.

Result: There were four girls and two boys. The average age was 8±3, 93 years with extremes of 3 and 15 years. Ten knees were involved. The webbing was knee-ankle- leg (7 cases) and hamstring leg (3 cases). The march was limping (2 cases) or lap (4 cases). Associated malformations were identified, sacral agenesis of vertebrae (3 cases), hypoplasia of the labia majora (2 cases) and incontinence of urine (1 case), bilateral cryptorchidism (3 cases), orofacial anomalies (3) and clubfoot (5). Surgery which was followed by equipment, were musculo-cutaneous plasty in the popliteal fossa (3 cases) or a leg amputation (4 cases).

Conclusion: The popliteal syndrome is the most important malformation of SPP, purveyor of disability, requiring screening and early orthopedic support that would limit the indication for amputation.

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Introduction

Popliteal pterygium syndrome is a rare disease with autosomal dominance. Its incidence is approximately 1 / 300,000 live births. It was first described by Trelat in 1869. The term "popliteal pterygium syndrome" was coined by Gorlin et al. in 1968 on the basis of the most unusual anomaly, the popliteal pterygium, a tissue filling of the posterior aspect of the knee limiting extension and handicapping bipedal walking [1-4]. The clinical features of this syndrome are highly variable and show different associations of abnormalities such as craniofacial, genitourinary and extremity abnormalities [5]. Its multidisciplinary treatment involves long and difficult reconstruction surgery for craniofacial, genitourinary and orthopedic anomalies [5].

Patients and Method

This retrospective and descriptive study covered the period from January 2006 to December 2015 (10 years) in the Pediatric Surgery department of the Cotonou Lagoon Mother and Child National University Hospital Center. It concerned children, aged from 0 to 15 years, received in consultation or hospitalized and taken care of

for popliteal pterygium. Sociodemographic data (frequency, age, sex; clinically, gestational history, walk, popliteal extension of the pterygium, strain angle of the fessum, associated malformations) had been studied as well as para clinical, therapeutic and progressive data over an average follow-up of 6 years.

The limited financial means of our patients hampered the exhaustive exploration of certain associated lesions. The patients had been operated on as part of the charitable surgical missions. Also, during the study period, magnetic resonance imaging and scintigraphy were not available in Benin.

Results

Epidemiological Aspects

In 10 years, 06 cases of popliteal pterygium were identified, either an annual incidence of 0.6. The sex ratio was 0.5. The mean age was 8+/- 3,93 years (range, 3-15 years). Gestational anamnesis was unremarkable in most patients. The mother and the brother of Patient 5 were carriers of arthrogyposis.

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Clinical and Therapeutic Aspects

These detailed aspects are shown in Table I.

In the 6 patients, ten knees were affected. The webbing was cruro-leg (7 cases) (Picture 1A) and ischio-leg (3 cases) (Picture 1A). Walking was limping (2 cases) in the unilateral involvement or on the knees (4 cases) in the bilateral involvement.

The average knee extension angle was $55^{\circ} \pm 17.63^{\circ}$ ranging between the extremes of 30° and 80° . The associated malformations identified were, sacral dysplasia (3 cases) (Picture 1B), hypoplasia of the labia majora (2 cases) and urinary incontinence (1 case), cryptorchidism (3 cases) (Picture 2B), and club feet (5 cases). Oral and facial abnormalities consisted of poor dentition (3 cases), a labial groove (3) and a velar cleft (1). All of the patients had normal growth and intellectual development. Three of them were in school.

The surgical interventions followed by equipment were either a musculo-cutaneous plasty at the level of the popliteal fossa (3 cases) (Picture 2C and 2D) or an amputation of the leg (4 cases) (Picture 1C and 1D). Parents of patients 5 and 6 refused the proposed amputation. Regarding the associated malformations, only the management of urinary incontinence had been carried out, by the creation of a continent bladder according to Mitrofanoff technique.

Table I: Epidemiological, clinical et therapeutical characteristics of patients

	Age	Sex	Web	Associated malformations	Treatment
Patient 1	3 years	F	Left ischio-stub (45°)	Lymphoedema of the affected foot Oral and facial abnormalities	Z-Plasty Iterative Plasters Equipment
Patient 2	10 years	M	Bilateral ischio-stub (30°)	Bilateral club foot Syndactyly Oral and facial abnormalities Bilateral Cryptorchidism Sacral dysplasia	Z-Plasty Iterative Plasters Equipment
Patient3	15 years	F	Bilatéral Cruro leggings (60°)	Hypoplasia of the labia majora Urinary Incontinence Coxal Arthrosis Sacral dysplasia Agenesis of the toes	Amputation Equipement
Patient 4	8 years	F	Bilateral Cruro- leg (60°)	Club foot Hypoplasia of the labia majora Oral and facial abnormalities Sacral dysplasia	Amputation Equipment
Patient 5	8 years	F	Bilateral Cruro-leg (80°)	Bilatéral club foot Oral and facial abnormalities	No
Patient 6	7 years	M	Left Cruro-leg (45°)	Oral and facial abnormalities Left cryptorchidism	No



Picture 1A: bilatéral cruro-legging web



Picture 2A: bilatéral ischio-stub web



Picture 1B: sacral dysplasia



Picture 2B: bilateral cryptorchidism



Picture 1C: amputated legs and malformed feet

Picture 2C: Z-plasty and nerve root exposure



Picture 1D: Patient 3 after equipment

Picture 2D: Patient 2 after equipment

Evolutionary Aspects

Patient 1 has moved and is lost to follow-up. Patient 2 presented with a scar band which reduced the acquired extension of the knees after 5 years (Picture 2E). His knees had retracted; he needed surgery and braces again. This could not be achieved due to lack of financial means. He dropped out of college and currently uses a wheelchair. Patient 3 gets around with her equipment, also uses a wheelchair if needed. She empties her continent bladder by probing. She reached a pre-university level and had a normal motherhood. Patient 4 travels with her device and continues her education normally.



Picture 2 E: Patient 2 with retractable clamps

Discussion

The monocentric nature of this study does not provide national statistics on PPS in Benin. However, this study has the advantage of being the first in our country. It therefore constitutes a starting point for further studies.

Epidemiological Aspects

The exact annual prevalence and incidence are unknown, but a prevalence around 1 / 300,000 individuals has been suggested for popliteal pterygium syndrome [5]. In our study, an annual incidence of 0.6 cases was found. This incidence could be higher if the care was financed and if the social context did not favor infanticide [6].

In the literature, we do not find a predominance of sex (sex ratio at 1), on the contrary, there was a strong female predominance in our study [7]. This could be explained by the fact that parents do not support the social gravity of the handicap caused by the malformation when it comes to girls. As proof, patient 6 with the malformation on only one knee, using a crutch to move, did not return for treatment and would probably have adapted to his condition.

The average age of patients at admission was high and is believed to be linked to poverty and lack of funding for care. Charitable missions had provided opportunities for screening and treatment. This is certainly why the literature review in sub-Saharan Africa remains poor in confirmed cases of popliteal pterygium syndrome [8].

Clinical and Therapeutic Aspects

Popliteal pterygium syndrome is an autosomal dominant congenital malformation. It is due to a mutation of the IRF6 gene in its 1q32.2-q32.3 region involved in the formation of epithelial and connective tissue. It has Van der Woude syndrome as an allelic variant [9]. This malformation can be diagnosed during prenatal period, especially for high-risk pregnancies, by molecular analysis after amniocentesis or chorionic villus sampling, if the mutation has been identified within the family [9]. In our context, the case of patient 4 seems to evoke a family transmission since the mother and the younger brother presented forms of arthrogyposis. The refusal of treatment did not allow further investigation. In addition, the existing technical platform does not allow easy access to these explorations but also, the gestational history was poor overall, since pregnancies were often poorly monitored.

Popliteal pterygium syndrome consists of the following abnormalities: cleft lip and / or palate, pits or sinuses in the lower lip, popliteal pterygium, bifid scrotum, cryptorchidism in boys, hypoplasia of the labia majora, syndactyly of the fingers and / or toes and skin abnormalities around the nails. Most patients have a pyramid-shaped skin fold on the nail of the hallux. Some have missing teeth. Other occasional manifestations include threadlike synechia connecting the upper and lower jaws (syngnathia) or the upper and lower eyelids (ankyloblepharon) and club feet [1-4, 9]. The abnormalities found in our study were popliteal pterygium, cryptorchidism, hypoplasia of the labia majora with urinary incontinence, club feet, syndactyly, and skin folds on the hallux and agenesis of the sacral vertebrae. The oral and facial abnormalities on the face of patient 2 appear suggestive of ankyloblepharon and pipefish. An x-ray was not

taken for documentation. Growth and intellectual development were normal in all of our patients as described by some authors [10]. Popliteal pterygium can also be found in other syndromes such as multiple pterygium syndrome and congenital multiple arthrogyposis [10]. These cases have not been identified in our patients.

The management of popliteal pterygium syndrome is multidisciplinary. There are several methods of treating popliteal webs consecutive to pterygium. According to some authors, surgical treatment is preferable to conservative treatment, including repetitive pull-ups which are not satisfactory [11]. Among the surgical means, lengthening of soft tissues such as skin, muscles and ligaments using resection of fibrous bands and Z-plasty were the treatments of choice, as in 3 of the cases in our series [11-13]. Sometimes a femoral extension osteotomy, shortening of the femur or knee arthrodesis is done. In cases of severe stiffness and hypoplasia of the leg bones, amputation becomes an option [14]. This was the mode of treatment offered to 4 of our patients. Two accepted while two refused. The use of amputation can be explained by the delay in consultation and therefore in support. They had all benefited from a second stage fitting. This is also justified by the social context of poverty where more surgeries require more money with uncertain results like patient 2, for whom a first-line amputation would have given more comfort in life and the chance to correct associated malformations such as bilateral cryptorchidism.

An Ilizarov External Fixator is commonly used for correcting shortening or deformity. It is also used for the treatment of stiff joints. According to Gillen et al, the device allows an extension of 0 °, but a contracture of 15-30 ° could occur after surgery. This rebound phenomenon is a major obstacle in the treatment of popliteal pterygium. In our study, none of our patients was treated with an external fixator [15].

Conclusion

Popliteal pterygium syndrome is a rare condition involving several malformations. It is a provider of multiple infirmities. Its management is multidisciplinary. The treatment of the resulting popliteal web is long, heavy, painful and very expensive. Early detection and management would limit the indication for amputation.

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