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Case Report

A 10-year-old Bangladeshi Girl with Popliteal Pterygium Syndrome: A Case Report

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Abstract:

Popliteal pterygium syndrome (PPS) is a unique congenital malformation affecting the limbs, face, and genitalia, named based on the most distinctive feature, the popliteal pterygium. Affecting less than 0.1% of the population, only a few publications have been found on PPS. Despite having no cognitive or systemic dysfunction, the physical features of PPS makes it difficult for the patients to walk or maintain a normal lifestyle. Most of the anomalies in PPS are surgically treatable and therefore, early diagnosis and treatment allows a standard life for the affected.

In this report, we present a ten-year-old girl presented with bilateral popliteal pterygium, treated cleft lip, an untreated complete cleft palate and hypoplastic labia majora. Her management approach is furthered discussed in the report.

Introduction

PPS is a rare autosomal dominant disorder first reported in 1869 with an incidence of 1 in 300,000 per live births[1]. This genetic disorder is caused by a heterozygous mutation in the interferon regulatory factor-6 (IRF6) gene on at chromosome 1q32.2[2]. As haploinsufficiency of IRF6 is associated with orofacial, cutaneous, musculoskeletal, and genitourinary developmental disruption, therefore, the cardinal features of PSS may include disorders in all or some of these systems[2]. Patients usually present with any combination of anomalies such as cleft lip and/or palate, lower lip pits or sinuses, syndactylies, pyramidal nail fold and a very distinctive popliteal pterygium or web[3]. Additionally, female patients present with hypoplastic labia majora and bifid scrotum and cryptorchidism may be observed in male patients. Other oral and musculoskeletal anomalies such as syngnathia, oral adhesions may also be found[1,4].

Approximately twenty cases of PSS have been published worldwide, none of which have been from Bangladesh. Therefore, the case described in this report will be the first to be published in Bangladesh. Two other cases from here were presented and are yet to be published.

Case summary

A 10-year-old girl presented with congenital longitudinal webbing across the posterior aspect of both lower limbs. The web extended from gluteal region to the heel on the right side and gluteal region to upper part of leg on the left side *(Figure 1A, 1B)*. A dense fibrous cord was also observed on the posterior portion of both legs along with unilateral syndactyly of right foot and wide gap between great toe and second toe of same foot *(Figure 1C).*

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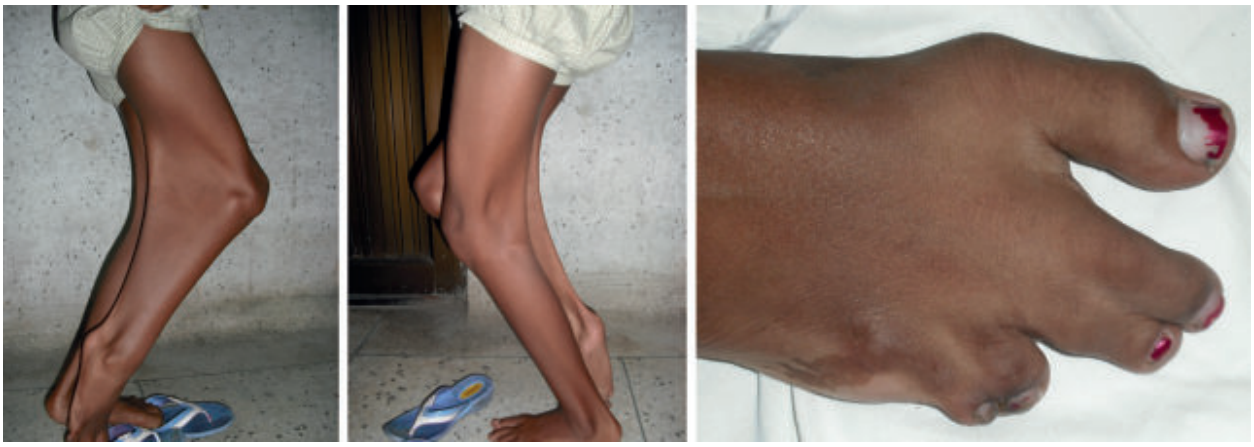

Figure 1: Popliteal pterygium on both lower limbs extending from the gluteal region to heel on the right (A) and gluteal region to the upper part of leg on the left (B). The right leg also demonstrated syndactyly and wide gap between great toe and second toe (C).

In addition to these skin webs, she had left sided complete cleft lip, which was repaired, complete cleft palate, pitting in the lower lip, hypertelorism (asymmetrical gap between the medial canthus of both eyes) *(Figure 2)*, bilateral hypoplastic labia majora, hypertrophied clitoris (Figure-3) and right sided equinism. Examination of the cardiovascular and respiratory system revealed no abnormality.

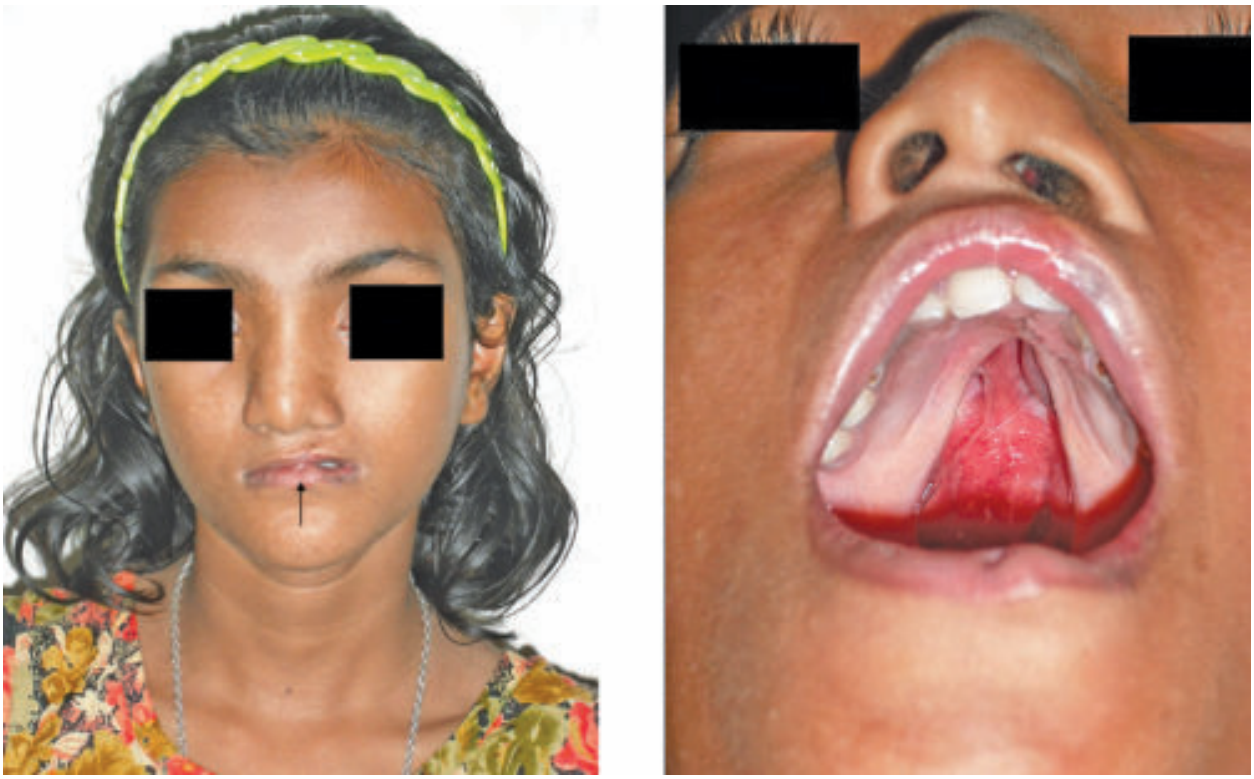

Figure 2: *The image on the left demonstrates the characteristic pitting in the lower lip (arrow), hypertelorism and a scar on the left side of upper lip from surgical repair of the cleft lip. The image on the right demonstrates the complete cleft palate.*

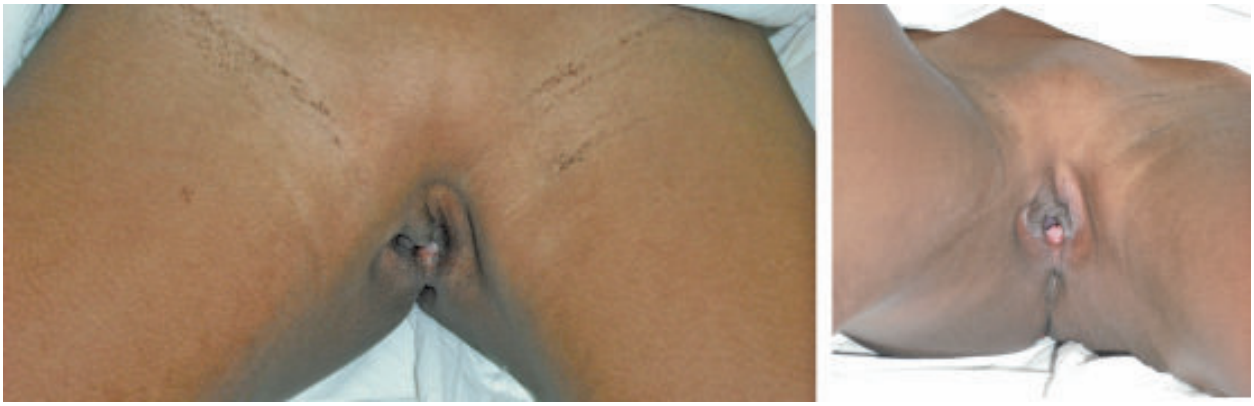

Figure 3: *Both images demonstrate the genital anomaly of the patient: bilateral hypoplastic labia majora and hypertrophied clitoris.*

Her mother also had cleft lip, cleft palate and pitting in the lower lip. She had an uncomplicated pregnancy and delivery when this girl was born. No other member of their family had a similar deformity.

Discussion

PPS, also known as facio-genitopopliteal syndrome is an unusual malformation disorder affecting approximately 0.0003% of the worldwide population. Patients frequently present with lower lip pits with cleft lip and/or palate, popliteal pterygium, genital, and musculoskeletal anomalies. Any three of the anomalies mentioned can be considered as the minimal diagnostic criteria for diagnosing PPS[5].

The most frequent anomaly is cleft palate with or without cleft lip (present in 97% cases), however, the most characteristic feature of the disorder is flexion contracture of knee and due to popliteal pterygium or web (present in 58% cases)[6,7]. The web most commonly extends from the ischium to the heel[7]. This leads to shortening of sciatic nerve and popliteal vessels, causing a characteristic equine gait. Additionally, 50% cases presented with syndactyly and 37% presented with genital anomalies. Interestingly, our patient presented with all the above-mentioned features that led us to clinically diagnose her as a case of PPS. Patients may also present with other features such as, ankyloblepharon, talipes, syngnathia, oral adhesions, spina bifida occulta, bifid ribs and short sternum[3]. Despite having numerous physical disabilities, growth and intelligence is usually unaffected in patients with PPS[8].

Another interesting genetic disorder that often presents with similar features as PPS is Van der Woude syndrome. Van der Woude syndrome comprises of approximately 2% of all cleft patients and often the most defined syndrome is lower lip pit with or without cleft lip and/or palate. Additional association of features with PPS are hypodontia and a unique big tooth in a small premaxilla[9]. Despite having similar anomalies, one of the biggest differences between the two conditions is the presence of a popliteal pterygium. Interestingly, IRF6 gene dysfunction is responsible for both PPS and Van der Woude syndrome. IRF6 gene is involved in forming epithelial and connective tissue and therefore, mutation in these genes lead to the multiple connective tissue related anomalies in both syndromes. In PPS, there is often missense mutation of the IRF6 gene in chromosome one whereas, in Van der Woude syndrome, there is deletion and mutations of the same gene[10]. Molecular analysis is often used for confirmatory diagnosis of by analyzing the type of mutation in IRF6 gene alongside the clinical features. Despite having similar features, the treatment protocols of both syndromes may differ as Van der Woude syndrome lacks the popliteal web that is specific to PPS. Our case could also be differentiated from multiple pterygium syndrome by the absence of multiple joint involvement, as in multiple pterygium syndrome, the pterygium may affect the axilla, elbow and interphalangeal joints[11]. Another congenital anomaly, namely, Arthrogryposis multiplex congenita can also be ruled out as it involves symmetrical joint deformity and movement restriction due to amyotrophy with loss of skin creases[11].

The treatment approach to PPS is almost always surgical, whether it is correction of the popliteal web,

orofacial anomalies, genitourinary anomalies, or musculoskeletal anomalies. Conservative treatment such as serial casting or traction of popliteal web have been proven to be substandard. Whereas surgical excision of the fibrous band along with lengthening of the skin, muscle and ligaments and multiple Z-plasties have shown to be more affective long-term[7]. Rarely, the sciatic nerve may be attached to the fibrous cord in which case, nerve grafting must also be considered following lengthening of soft tissue. Additionally, postoperative physiotherapy is also essential to ensure lasting benefits. Often when the primary surgery cannot be performed such as in severe sciatic nerve adhesion, secondary surgeries such as femoral extension osteotomy, femoral shortening or amputation may be required[11]. For our patient, Z-plasty and excision of the fibrous band along with the border of the pterygia was performed. The procedure was followed by application of dynamic split which allowed progressive stretching of the tissue. Additionally, our patient had left sided complete cleft lip, which was repaired, however, the complete cleft palate was not. Therefore, the next management plan is to repair the complete cleft lip as soon as she recovers from her limb surgery.

Conclusion

We report the first ever published case of PPS in Bangladesh in this report. Our patient presented with features typical of the syndrome and was managed through surgical intervention followed by dynamic split which allowing progressive stretching of the tissue. As PPS is a genetic disorder, therefore, proper antenatal genetic checkups in susceptible women may prepare the parents and patients for early diagnosis and management leading to a better and long-lasting outcome.

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