Surgical Management of Persistent Fetal Vasculature

Persistan Fetal Damar Sendromunda Cerrahi Yaklaşım

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ABSTRACT

Persistent fetal vasculature (PFV), also known as persistent hyperplastic primary vitreous, is a congenital developmental malformation of the eye, caused by the failure of involution of the primary vitreous and hyaloid vasculature. The severity of the disease may vary depending on the affected intraocular structures and anatomical variations, and the broad spectrum of clinical presentations makes PFV challenging for surgical management. The proper selection of surgical techniques is critical to prevent complications and achieve favorable outcomes and highly depends on knowledge of the type, location, and extent of the disease. The aim of this work is to describe the clinical, anatomical features and variations of PFV, as well as to review the principles and pearls for successful surgical management in the current literature and our clinical experience.

Key Words: Persistent fetal vasculature; PFV; Persistent hyperplastic primary vitreous; anterior retinal elongation; surgical management.

ÖZ

Persistan hiperplastik primer vitreus olarak da bilinen persistan fetal damar sendromu (PFD), embriyolojik primer vitreus ve hyaloid damarlarının gerilemesinde yetersizlik sonucu ortaya çıkan konjenital gelişimsel bir anomalidir. Hastalığın şiddeti, etkilenen göz içi yapılara ve anatomik varyasyonlara bağlı olarak değişkenlik gösterebilmektedir ve klinik prezentasyondaki bu çeşitlilik cerrahiyi zorlaştırmaktadır. PFD tipi, yerleşimi ve uzanımının bilinerek uygun cerrahi tekniklerin seçimi, komplikasyonları önlemek ve olumlu sonuçlar elde etmek için kritik öneme sahiptir. Bu çalışmanın amacı, güncel literatür ve klinik deneyimlerimizin ışığında, PFD'nin klinik ve anatomik özelliklerini ve varyasyonları tanımlamanın yanı sıra başarılı cerrahi tedavi için uygulanması gereken ilkeleri gözden geçirmektir.

Anahtar Kelimeler: Persistan fetal damarlanma; PFD; Persistan hiperplastik primer vitreus; retinanın öne uzanımı; cerrahi yönetim.

INTRODUCTION

Persistent fetal vasculature (PFV), previously known as persistent hyperplastic primary vitreous, is a congenital developmental abnormality caused by failure of regression of the primary vitreous and hyaloid vasculature.[1] The disease typically presents unilaterally in full-term infants without associated systemic findings, but rarely it may be in conjunction with systemic syndromes such as Norrie's disease,[2] Walker- Warburg syndrome,[3] Aicardi syndrome,[4] trisomy 13, 15, 18[1, 5] and may be bilateral. Overall, bilateral cases account for less than 10% of the cases.[6] The exact etiology of PFV is not known and most of the cases are sporadic.[6] Nevertheless, rare cases of autosomal dominant and autosomal recessive inheritance have been reported related to the genes NDP and ATOH7, respectively.[7, 8]

CLINICAL FINDINGS

PFV is a spectrum of disease that involves the remnants of the fetal hyaloid system, extending from the optic disc to the lens to varying degrees. It can be classified as anterior, posterior, and combined forms, according to the affected intraocular structures.

The patients with the purely anterior variety of PFV characteristically present with leukocoria very early in life. The white pupil is often a result of the fibrovascular

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retrolental membrane alone or in combination with a cataractous lens. The eye is usually microphthalmic; however, this can be subtle in some cases. The ciliary processes are generally elongated and centrally dragged into the fibrovascular membrane. Prominent and dilated radial iris vessels and hairpin loops may be observed in some cases. A shallow anterior chamber due to anterior displacement of the iris- lens diaphragm, as well as extensive posterior synechiae and peripheral anterior synechiae may be seen secondary to fibrovascular tissue in ectopic locations and often progress to secondary angle-closure glaucoma. In contrast to common observation of microphthalmos, patients with this complication may present with normal-sized eyes or buphthalmos secondary to glaucoma. Posterior pole, optic nerve and macula are totally normal in the purely anterior forms with no evidence of a retinal fold. However, a remnant of a thin hyaloid stalk may be found extending from the optic nerve head with the anterior end lying freely in the vitreous gel.

Posterior subtype consists of an elevated vitreous membrane and a stalk from the optic nerve, hypoplastic or dysplastic optic nerve, retinal fold or tractional detachment. The eye is usually microphthalmic, but can be of normal size. The lens is often clear; however, it may become cataractous with time if the vessels from the membrane grow forward enough to enter the lens through the posterior capsule. Nevertheless, most of the cases exhibit features of both anterior and posterior forms. Moreover, PFV can also occur in conjunction with other ocular abnormalities, including morning glory disc anomaly,[9] Axenfeld-Rieger syndrome,[10] Peter's anomaly, uveal coloboma, macular hypoplasia, and microcornea.[1] Variety of such presentations makes PFV challenging for surgical management.[6]

DIAGNOSIS

Direct visualization of any component of the persistent vascular remnants is the best means for diagnosis. B-scan ultrasonography can be extremely useful to assist in diagnosis, especially if there is a limited or absent view of the fundus. Besides showing the vitreous stalk from the posterior pole to the lens, it can provide additional information concerning the axial length, status of the lens, presence of RD.

Since the most commonly recognized clinical manifestation of PFV is leucocoria, retinoblastoma must be on the differential diagnosis for PFV although it is very rarely associated with microphthalmia. B-scan ultrasonography and computed tomographic scanning can be exceptionally helpful in the case of a diagnostic challenge. Calcification seen on imaging is suggestive of malignancy; however, due to the possibility of increasing the risk of malignancy in children with this form of radiation, an MRI might be a superior alternative.[6] Fluorescein angiography is another ancillary test that can delineate abnormal vasculature such as radially oriented iridohyaloid vessels or vessels forming a "brittle-star" configuration.

TREATMENT

A wide spectrum of presentation translates into a similarly wide range of treatment options and potential outcomes. At one extreme end, an eye may have minimal sequelae from the fetal vascular structures such as a Bergmeister's papilla, a Mittendorf dot, muscae volitantes, or a persistent pupillary loop. As Goldberg[1] indicated, many minimally affected eyes, such as those with either normal visual function or clear visual axis and lack of progressive anatomic changes, run an uncomplicated course and remain stable without surgical treatment. Surgery may also be avoided for those at the other extreme end of the spectrum of severity, especially when PFV is associated with systemic syndromes and the retina or the optic nerve is severely undifferentiated, or dysfunctional that the surgery would be clearly not effective. Yet for most of the cases with PFV, surgery is the only option to salvage the eye and to improve vision.[11-13]

The main goals of surgery for PFV are to clear the media to prevent amblyopia and relieve tractional forces to prevent complications such as retinal detachment (RD), glaucoma and phthisis. The persistent hyaloidal stalk is attached anteriorly to the posterior lens capsule and posteriorly to the vascular bundle of the optic nerve head. This stalk remains the same and rigid in the growing eye and exerts tractional forces radially in all directions. If left untreated, anterior traction can create tractional RD, retinal dysplasia, and optic nerve hypoplasia. Posterior traction on the lens capsule may cause posterior lenticonus and circumferential traction on the vitreous base can create combined tractional and rhegmatogenous RD, vitreous hemorrhage, and ciliary body detachment with hypotony and phthisis.[6] By releasing the connections of the rigid hyaloidal stalk, the eye has the opportunity to grow and acceptable anatomic and functional results can be achieved.[6, 13]

Reported outcomes of surgery for PFV vary considerably depending on the severity and type of the disease. It is assumed that purely anterior PFV cases generally have a good visual potential, whereas patients with posterior PFV often have poor prognosis because of coexisting retinal and optic nerve abnormalities. However, it has been demonstrated that earlier diagnosis and intervention within the early critical periods of visual development improve outcomes even for posterior cases.[12, 14, 15] Bosjolie et al.[15] emphasized that the retina has a period of plasticity in which surgical intervention should take place. In their series, 10 out of 11 patients, who underwent surgery before 13 months (average age, 4 months) for posterior PFV achieved significant reversal of retinal dragging, along with 20/800 or better vision. Similarly, in a series of 44 eyes who received surgery for all types of PFV, Karaçorlu et al.[16] reported that the average surgical age was 4 months for the 11 eyes with

20/400 or better vision, and 7 months for the 9 eyes with final vision of light perception or no-light perception, suggesting that earlier intervention may yield superior results. However, the results were poorer for the eyes with severe posterior involvement who underwent vitrectomy at an average age of 14 months, close to the critical period defined by Bosjolie et al;[15] out of 8 eyes, only 3 had attachment of the retina and 1 eye achieved a measurable vision of 20/400. Recently, we published our series of 29 eyes with anterior, posterior and combined PFV and reported the surgical outcomes and complications.[17] We observed that among the 15 eyes with posterior involvement, the majority had postoperative attachment of the retina (80%) and more than half of the patients achieved a final vision better than fix-follow (60%) (Fig.1). Although the eyes with purely anterior PFV had a higher percentage of measurable visual acuity of 20/400 or better, the results suggested that even patients with posterior components may achieve useful vision after surgery in conjunction with postoperative amblyopia treatment.

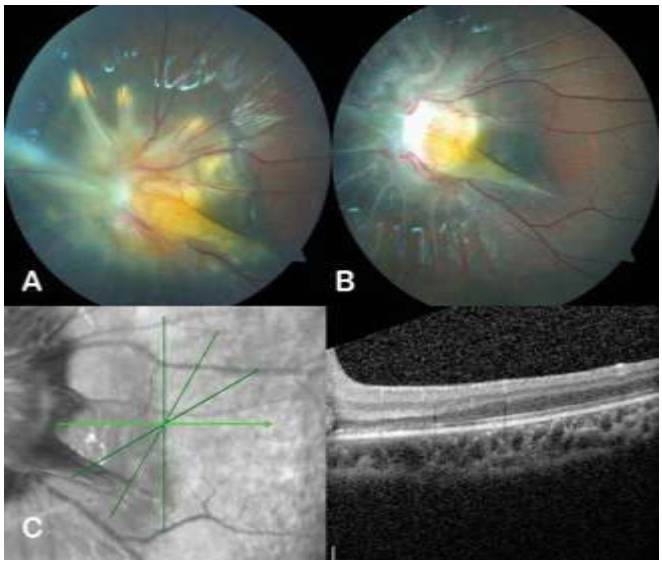

Figure 1. *Preoperative (A) and postoperative (B) fundus photos and postoperative OCT image (C) of a 4-year-old girl presented with strabismus; purely posterior PFV was found on initial examination with 10 PD XT. After lens sparing vitrectomy, tractions were relieved, VA improved to counting fingers at 2 meters and strabismus improved remarkably at 8 months.*

SURGICAL TECHNIQUES AND ANATOMICAL VARIATIONS

Although indications for surgical treatment have expanded and surgery has become more effective and safer with the advancement of vitreoretinal surgical techniques and disease knowledge over the years, development of complications such as retinal tears, RD, vitreous hemorrhage and pupillary obliteration are still a concern; especially in the presence of anatomical anomalies, such complications tend to occur more and compromise the success of the surgery.[5, 11, 14, 18]

Two alternative vitrectomy techniques can be utilized: limbal/transpupillary approach anteriorly or pars plana/pars plicata approach posteriorly. Although posterior approach provides better view and focus at the level of preretinal space and more precise control of maneuvers, it carries a risk of inadvertent retinal damage during insertion of the trocars. Several studies[5, 11, 14, 17] previously reported that the retina was dragged into the pars plana / plicata region in some PFV cases. As a result, some surgeons advocate the routine use of limbal approach suggesting that surgical avoidance of the ciliary body would lessen the risk of puncturing or excising retinal tissue.

Haddad et al.[5] reported abnormal insertion of the retina, anterior to the normal anatomic position, in 30% of the enucleated eyes with PFV, with only some of them reaching the edges of the retrolental mass in 1978. However, this was an old histopathological study; therefore, clinical importance of detection and management of this finding was not provided. Federman and associates,[11] reported on 7 patients treated with surgery for PFV. At the beginning of each surgery, they tried to localize the pars plana with transillumination. However, this usually failed. Then the sclerotomies were made between 2.0 mm and 3.5 mm posterior to the limbus. In one case, a portion of peripheral retina prolapsed through the sclerotomy. In a second, two holes were made in the peripheral retina where the vitrectomy instruments had passed through the sclerotomies. In a third, a 200° retinal dialysis occurred. The authors stated that these complications were due to the fact that the retina inserted directly into the pars plicata in these cases. In another report reported by Stark et al,[14] a pars plana approach had been used in the only eye that was complicated with RD after surgery. Given this, the authors suggested the use of the limbal approach because they believed that pars plana was often narrow and that anterior retinal placement into the retrolental membrane was possible.

Our recent study[17] has indicated that this peripheral retinal anomaly may be a much more prevalent finding, and may be of prognostic importance. In a series of 29 eyes with PFV, we have documented that, in 82% of the anterior PFV cases, the peripheral retina was dragged anteriorly, replacing the pars plana totally in some parts, and elongating as finger-like extensions or sometimes circumferentially beyond the ora serrata to become continuous with the retrolental fibrovascular tissue (Fig.2). We did not encounter this finding in any of the combined cases; however, surprisingly, one case with posterior PFV had peripheral retinal anomaly. Because the retina was continuous with the fibrovascular tissue, three eyes were complicated with iatrogenic peripheral retinotomy secondary to the removal of retrolental fibrovascular tissue (Fig.3). Another two eyes, in which the peripheral part of the fibrovascular tissue was left on purpose during primary surgery, developed peripheral RD in the postoperative period due to contraction of the fibrovascular remnant

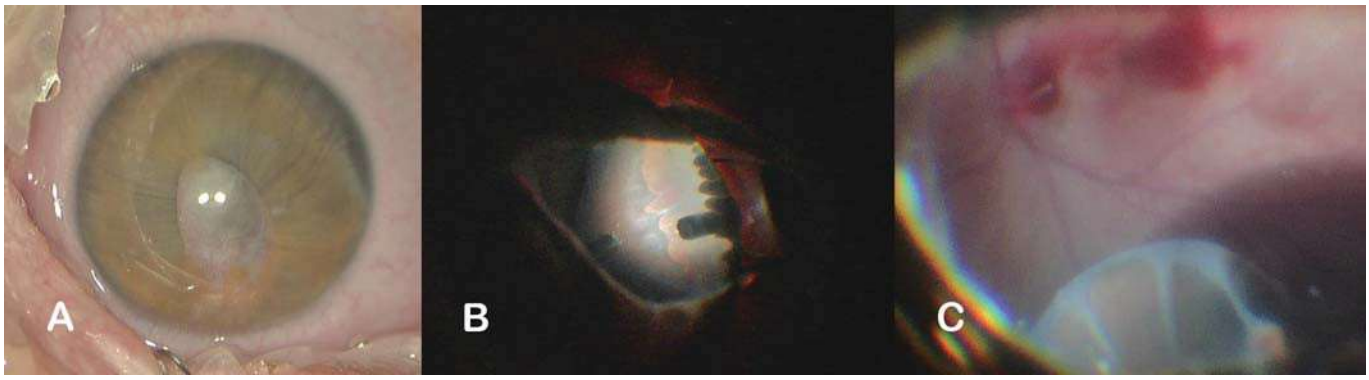

Figure 2. *An anterior PFV case with extensive form of fibrovascular membrane, elongated ciliary processes (B) and the peripheral retina elongating as finger-like extensions beyond the ora serrata (C).*

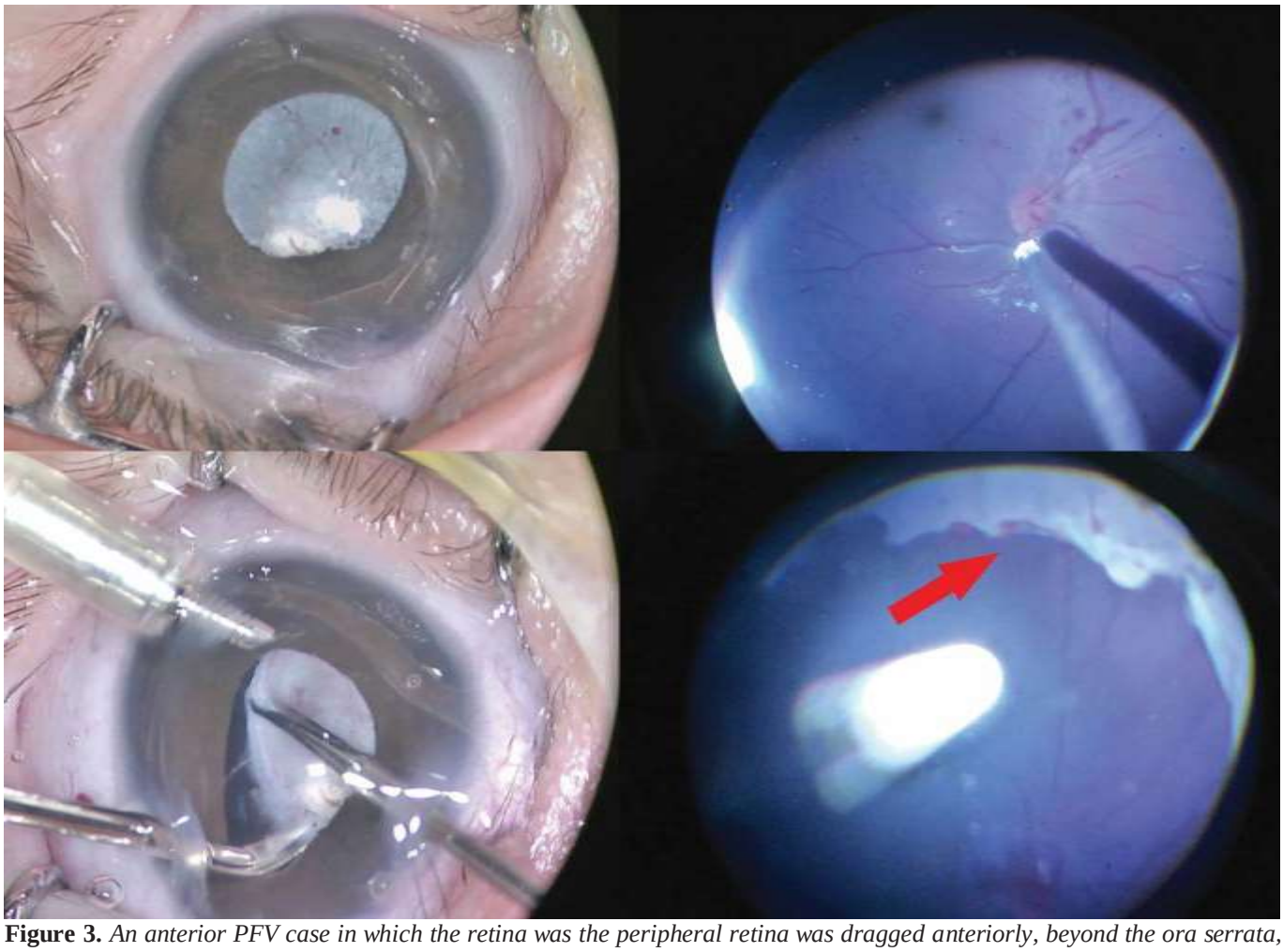

Figure 3. *An anterior PFV case in which the retina was the peripheral retina was dragged anteriorly, beyond the ora serrata, to become continuous with the retrolental fibrovascular tissue. When the fibrovascular tissue was cut, there was retina on the other side of the cut tissue which ended up with retinotomy.*

causing traction on the connected peripheral retina. In one of these cases, contraction of the fibrovascular remnant also caused pupillary obliteration with 360º synechia and angle-closure glaucoma (Fig.4). Noteworthy, these complications were found to be significantly associated with the extent of lens opacity. The results suggested that the eyes with more extensive retrolental opacities are more likely to develop complications and should be managed carefully.

We recommend careful investigation of the ora serrata-pars plicata region with indirect ophthalmoscopy, if possible, to be sure that there is no retinal elongation in the planned entry sites. In cases with an extensive fibrovascular membrane that obscures view, it would be safer to use limbal entries for the entire procedure or until an adequate view of the peripheral retina is achieved. Some clues also suggest the areas of retinal elongation which may guide the surgeon; nasal and

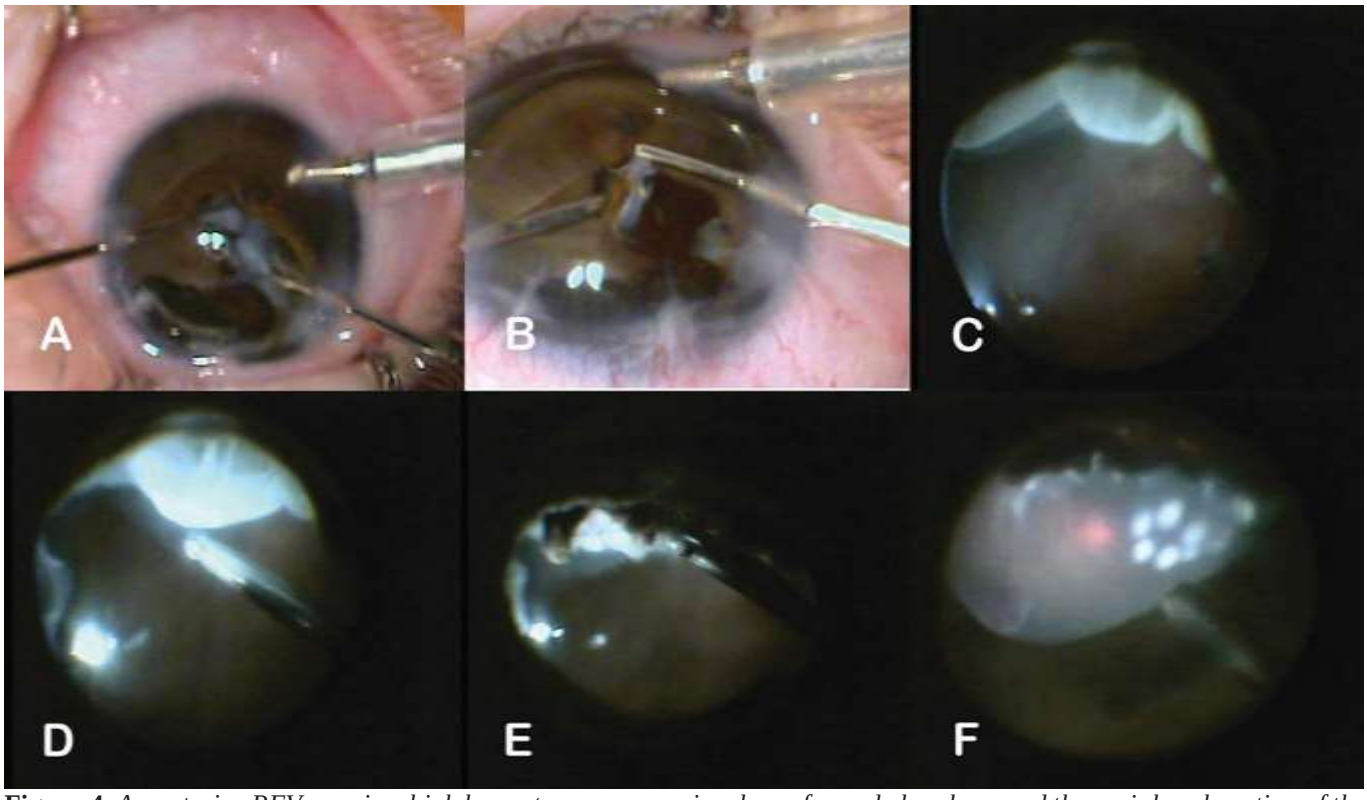

Figure 4. *An anterior PFV case in which lensectomy was previously performed elsewhere and the peripheral portion of the lens capsule was left in place for a secondary IOL implantation; pupillary aperture was obliterated by thick and tenacious membrane which is hard to separate (A-B). Peripheral retinal detachment is seen due to the traction of the membrane (C). Pupillary membrane was continuous with the detached retina in some parts (D-E). 360 degree retinotomy, photocoagulation and silicone tamponade enabled retinal reattachment (F).*

inferior parts contiguous to the localized pigmented or fibrovascular plaques are the places where elongated retina parts are most commonly located (Fig.5). Prophylactic laser photocoagulation can also be applied to these areas as necessary. Rarely, the cases that have localized fibrovascular tissue centrally located at the back of the lens may allow safe removal of the tissue and the preservation of the capsular bag for a secondary intraocular lens implantation. However,

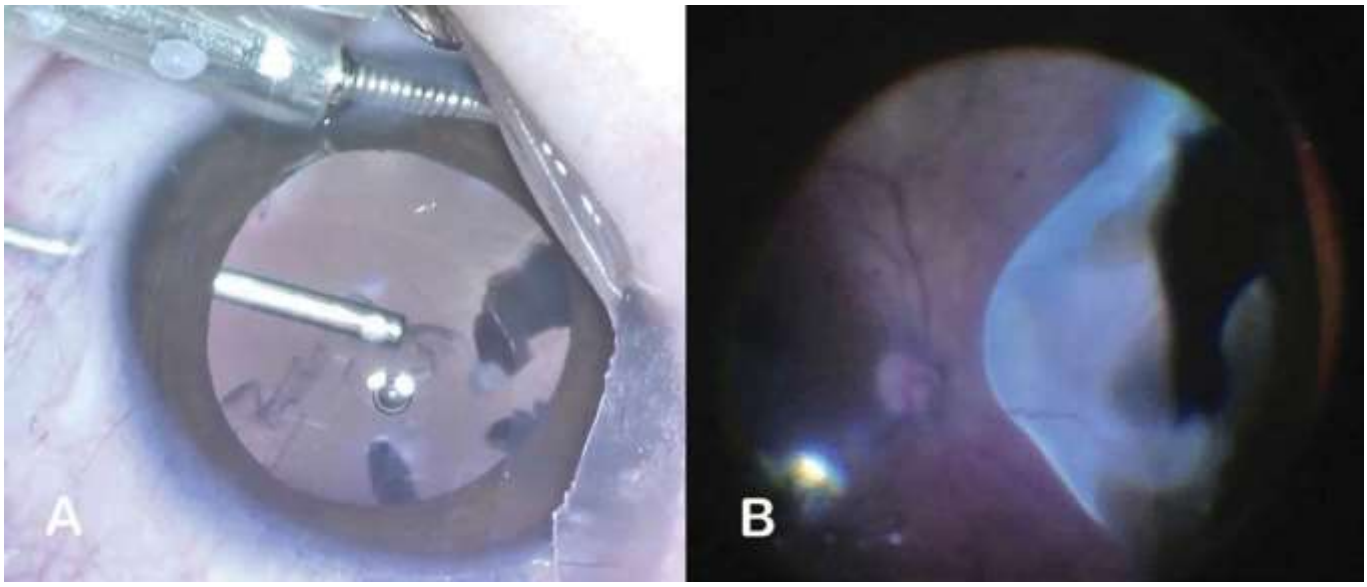

Figure 5. *Central cataract and eccentric nasal insertion of the fibrovascular stalk with an avascular pigmented membrane to the lens (A). Elongated retina, beyond the ora serrata, was continuous with the pigmented fibrotic tissue behind the lens inferonasally (B). This pigmented membrane behind the lens capsule was left in place since it was avascular and there was no risk of contraction in this particular case.*

for the cases that have more extensive and eccentric fibrous tissue which extends to the periphery of the back of the lens, the priority should be the complete removal of the retrolental fibrovascular tissue rather than preservation of the capsule to avoid potential complications. The surgeon must be ready to do the regular vitreoretinal surgery for rhegmatogenous RD if needed.

CONCLUSION

Surgical outcomes of PFV largely depend on the severity and type of the disease, early diagnosis and intervention, careful planning of the surgical technique, and close follow-up. Although purely anterior PFV cases have better visual potential, many cases with posterior malformations may achieve useful vision with proper care. Given the high rates of peripheral retinal anomalies, extra caution is warranted to avoid complications in anterior PFV cases who may otherwise have good visual potential. Refractive correction and effective occlusion of the fellow eye are as important as surgery to maximize visual potential. All in all, management of PFV cases requires meticulous care and follow-up that include early and proper surgery, family education and postoperative management of amblyopia.

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