Case Report

Retinitis Pigmentosa with a Bilateral Coats-Like Response – An Unusual Presentation

PJO – Official Journal of Ophthalmological Society of Pakistan

Sivaranjani[1], Deeksha Singh[2], Arun Jayanth[3]
Manavi D. Sindal[4]
[1-4]Aravind Eye Hospital, Cuddalore Main Road, Thavalakuppam, Pondicherry, India

ABSTRACT

Retinitis pigmentosa (RP) is an inherited retinal dystrophy that is usually bilateral with a female preponderance. We report a case of a 40-year-old male diagnosed with bilateral retinitis pigmentosa with coats like response. The diagnosis was confirmed based on clinical examination, typical findings on Fundus fluorescein angiography (FFA), Optical coherence tomography, B-scan and Indocyanine green angiography. Based on angiography findings, in both the eyes of our patient cryotherapy was done to the leaking telangiectatic vessels in the peripheral retina. On follow up, regression of the telangiectatic vessels was noted.

Key Words: Retinitis pigmentosa, coats disease, Mutation, CRB1 gene, Tumor.

How to Cite this Article: Sivaranjani, Singh D, Jayanth A, Sindal MD. Retinitis Pigmentosa with a Bilateral Coats-Like Response – An Unusual Presentation. 2024;40(3):342-346. **Doi:10.36351/pjo.v40i3.1773**

Correspondence: Sivaranjani
Aravind Eye Hospital, Cuddalore Main Road, Thavalakuppam, Pondicherry, India
Email: drsivaranjani.s@gmail.com

Received: July 11, 2023
Accepted: May 24, 2024

INTRODUCTION

Retinitis pigmentosa is an inherited retinal dystrophy that is bilateral. It mainly affects the rods in the peripheral retina, leading to progressive retinal photoreceptors degeneration. Approximately 1-5% of RP cases demonstrate coats like exudative retinopathy.[1] Inheritance is usually autosomal recessive. Mutation in the CRB1 gene was found to be the strongest association. Treatment modalities available for leaking telangiectatic vessels are laser photocoagulation and cryotherapy. Scleral buckling with SRF drainage is indicated if retinal detachment occurs as sequelae.

Case Description

A 40-year-old male presented with complaints of diminution of vision in both eyes (OU) since childhood, predominantly at night, progressively worsening during the past three months. His systemic and family history was non-contributory. No other family member was affected and there was no history of consanguineous marriage. On examination, his best-corrected visual acuity was 3/60 in the right eye and 1/60 in the left eye. His intraocular pressure was normal OU. Anterior segment examination was normal. Fundus examination of both eyes showed a pale disc with exudates noted over the disc (Figure 1).

Moreover, blood vessels were attenuated and retina showed bone spicule pigmentation with dilated telangiectatic vessels in inferotemporal quadrants and retinal exudation. Fresh and old vitreous hemorrhage was noted in both eyes. Full-field ERG revealed decreased b wave amplitude in scotopic response with extinguished waves in combined and photopic responses (Figure 2a, 2b).

Optical Coherence Tomography (OCT) revealed foveal thinning with vitreomacular traction (Figure 3a,3b). Fundus Fluorescein Angiography (FFA) and Indo Cyanine Green (ICG) angiography showed aneurysmal dilatation and telangiectatic retinal vessels with leakage (described as light bulbs) involving the temporal and inferior quadrants (Figure 4 a-h). B scan

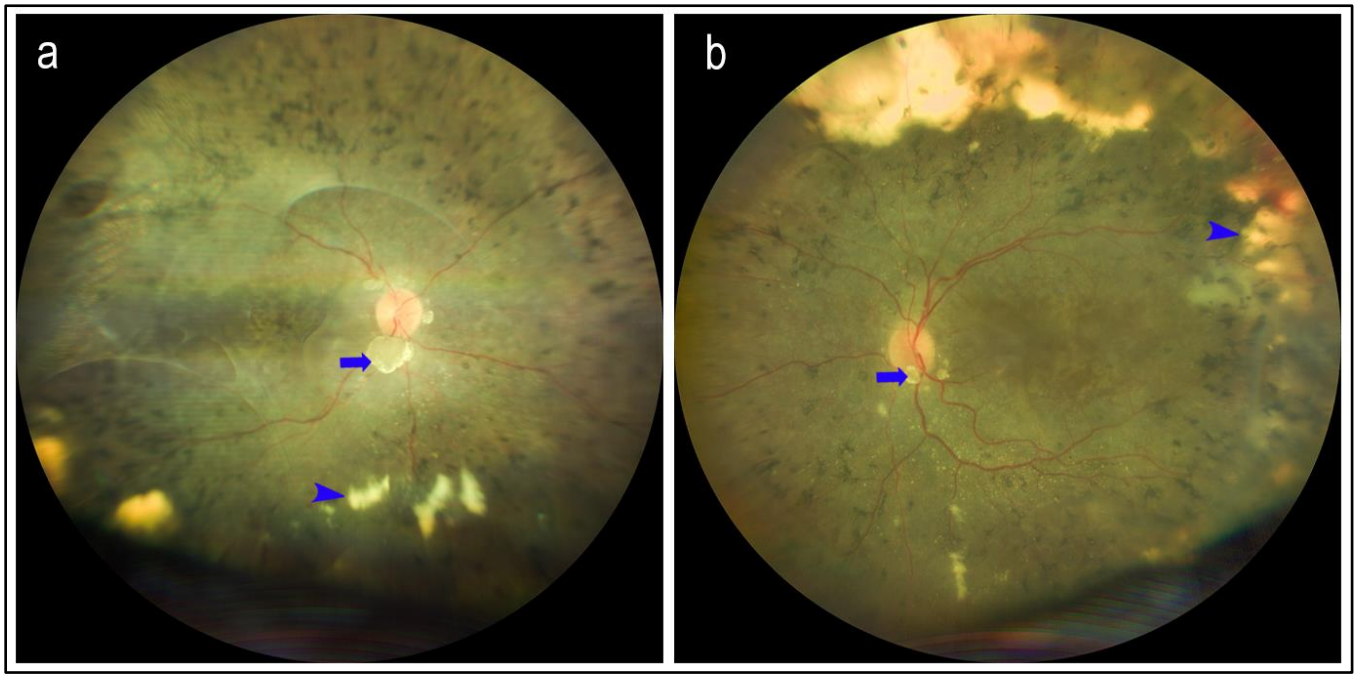

Figure1: Fundus photo of both eyes showing pale disc with exudates noted over the disc (arrow). Blood vessels were attenuated with bone spicule pigmentation in the mid periphery. Retinal exudates involving supero temporal and inferotemporal retina were noted (arrow head).

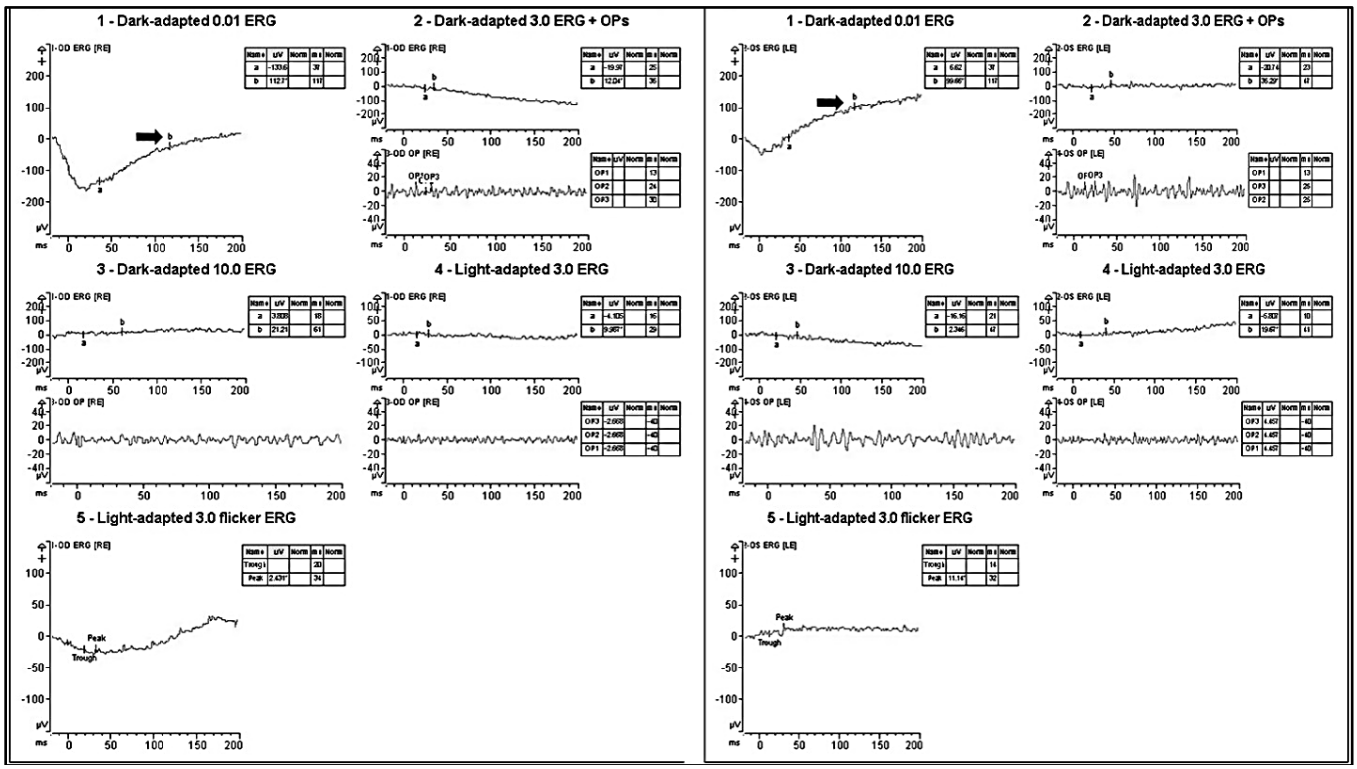

Figure 2: Full field electro retinogram of right eye (a)and left eye(b) shows severe reduction of b wave amplitude in dark adapted 0.01 ERG (arrow) with extinguished waves in dark adapted 3.0 ERG and light adapted 30 Hz flicker ERG.

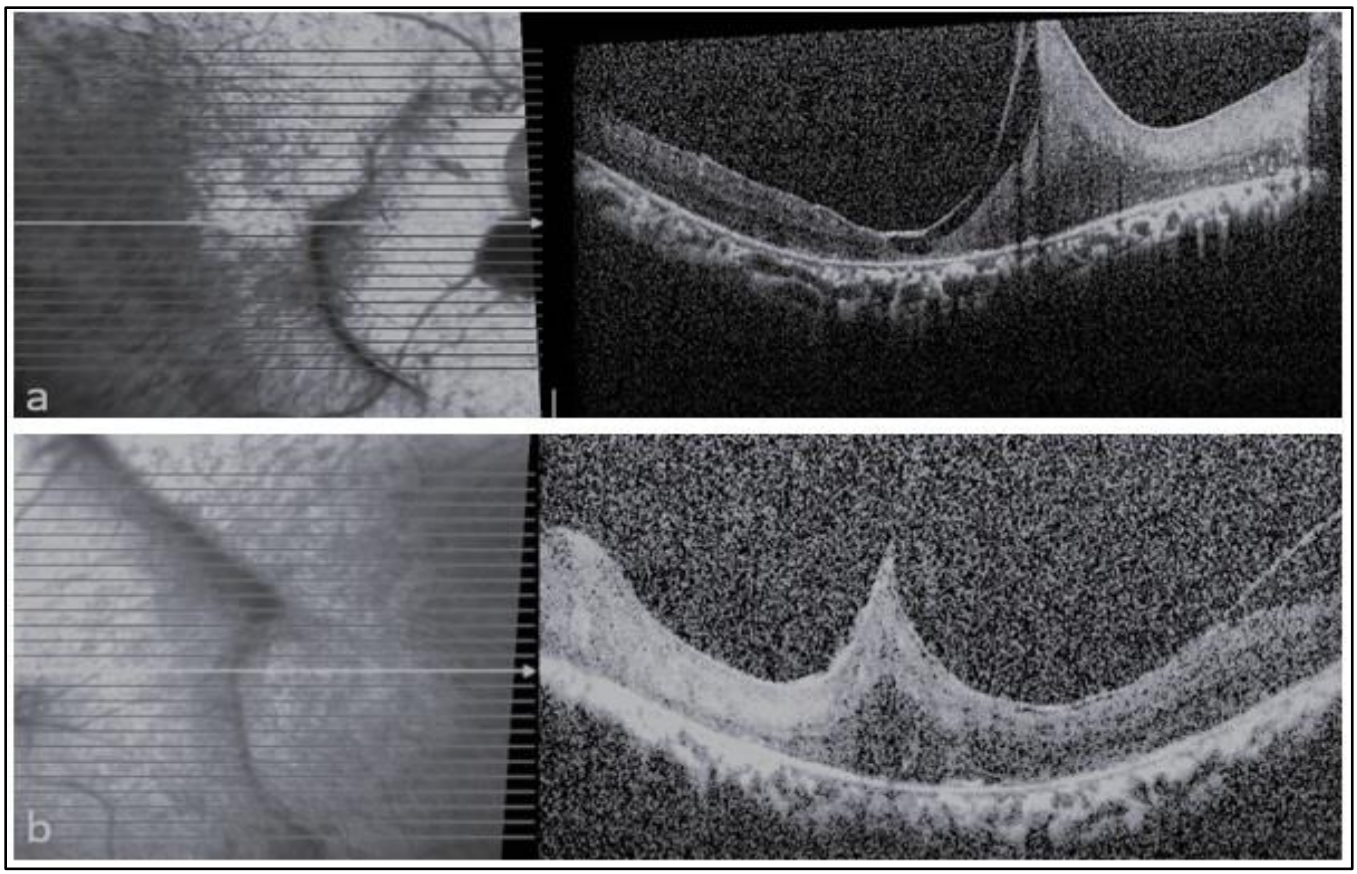

Figure 3: Optical Coherence Tomography (OCT) of right eye (a) macular cube 30°with horizontal line scan showing foveal thinning with vitreomacular traction and posterior hyaloid membrane. (b) left eye macular cube 30^0 with horizontal line scan showing vitreomacular traction involving the fovea.

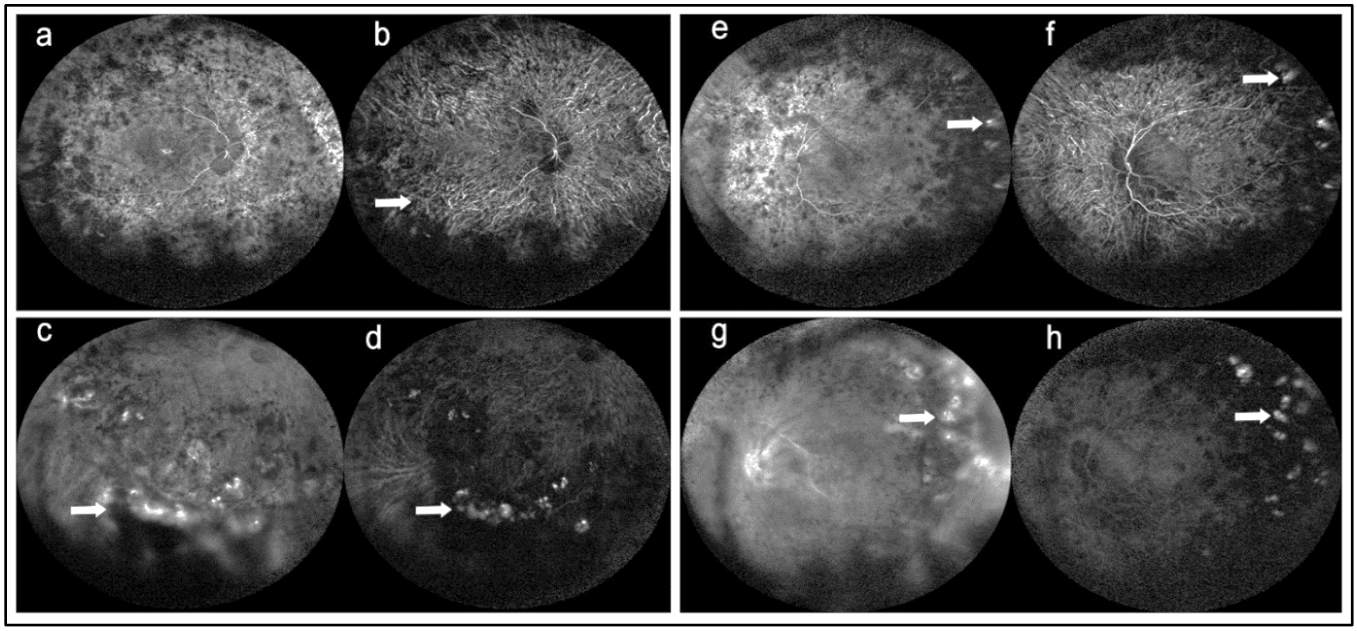

Figure 4: Right eye: a (FFA), b(ICG) shows hyper fluorescence due to telangiectatic vessels(arrow) early phase. c, d shows FFA and ICG of right eye in late phase with hyperfluorescence due to leakage of telangiectatic vessels described as light bulbs (arrow). Left eye: e (FFA), f (ICG) shows hyper fluorescence due to telangiectatic vessels as light bulbs(arrow) in early phase. G and h show FFA and ICG of left eye in late phase with hyper fluorescence due to leakage of telangiectatic vessels (arrow).

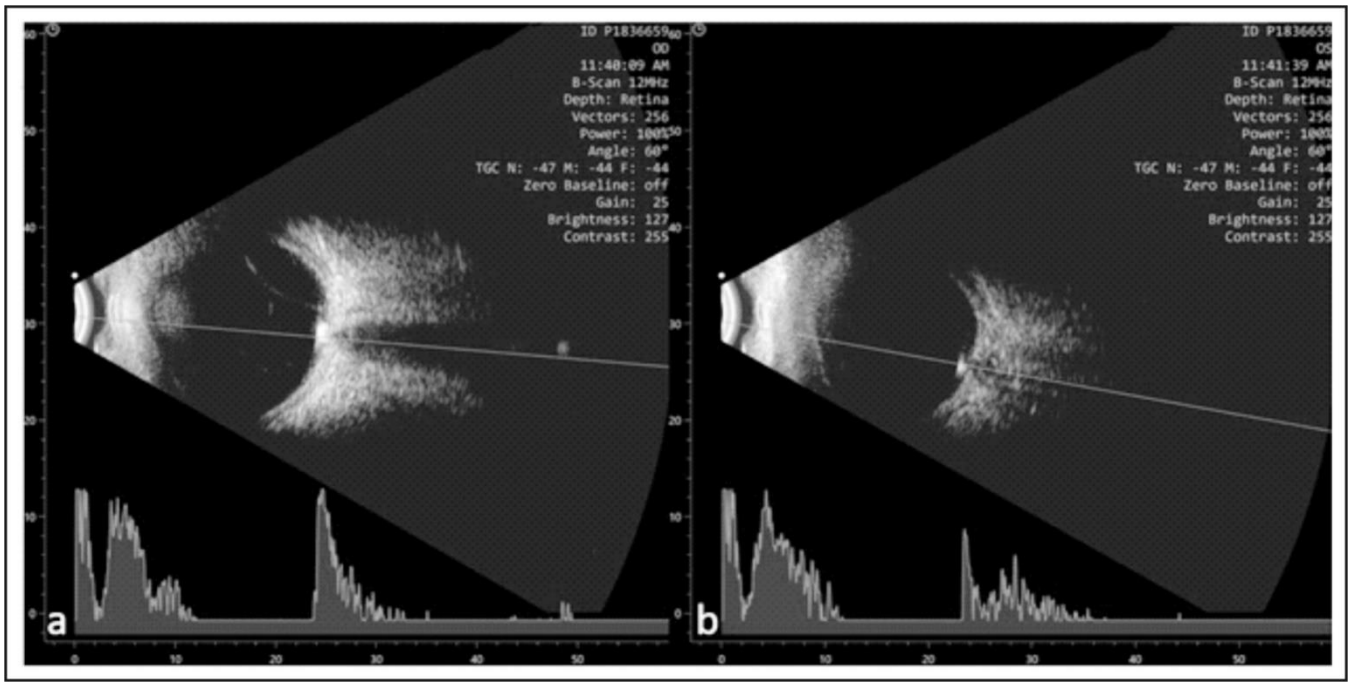

Figure 5: Right eye: (a), Left eye (b) shows B scan in axial plane with a hyper echoic lesion over the disc and corresponding high amplitude spikes in A scan due to chronic exudates over the disc.

of both eyes showed a hyperechoic lesion over the disc with acoustic shadowing due to chronic exudate over the disc (Figure 5a, 5b). A final diagnosis of retinitis pigmentosa with a coats-like response was made.

He underwent cryotherapy to the peripheral telangiectatic vessels in both eyes. At one month follow-up exudates showed regression with no new vitreous hemorrhage.

DISCUSSION

Zamorani was the first to describe association between RP and coats-like disease in 1956.[2] RP with coats-like disease manifests as telangiectasia or Vaso proliferative tumor with exudation. Mutations in the human homolog of the Drosophila crumbs gene (CRB 1) found the strongest association of RP with the coats-like response.[3] CRB1 gene plays a crucial role in cell polarity, cell-to-cell adhesion, photoreceptor morphogenesis, and retinal maturation. Blood-retinal barrier defect and degenerating photoreceptors lead to the accumulation of toxins which in turn cause vessels to leak.[4] This leads to hypoxia and development of telangiectasias and neovascularization. Other novel mutations are CNG 1, YULP 1, and RPGR.[5] We could not perform a genetic study on our patient as the patient was unwilling.

Another manifestation of RP with a coats-like response is the Vaso proliferative tumors (VPTs).[6] VPTs are pinkish-yellow dome-shaped masses in the peripheral retina with vascular and glial components.[6,7]VPTs are usually located in the peripheral retina in the inferotemporal quadrant. While primary VPTs are single unilateral lesions, secondary tumors are usually bilateral and multiple. In RP, coats-like response and VPTs are considered different stages of the same disease process.

Moreover, it is essential to differentiate between retinitis pigmentosa with a coats-like response and classic coat disease. RP with a coat-like response differs from classic coat disease by age, gender, eye involvement, retinal location and progression. RP with coats-like disease generally manifests in older age with female preponderance. However, our patient is a 40 years old male. RP with the coats-like illness is generally more progressive and located in inferior quadrants compared to classic coats disease, which manifests in young males with unilateral presentation. More commonly it involves the superior temporal retina and posterior location.

FFA-guided laser photocoagulation to the leaking telangiectatic vessels is recommended as the treatment of choice.[8] Cryotherapy is indicated in patients with dense exudates where laser is ineffective. Anti-VEGF

can be added if there is associated cystoid macular edema. Surgical intervention will be required if VPTs and RP with the coats-like disease are related to retinal detachment.

In conclusion, retinitis pigmentosa with a coats-like response is a rare entity that can be missed in the initial stage. Currently, no therapy stops or slows down the progression of the retinitis pigmentosa, so the visual prognosis is poor. Detailed peripheral fundus examination is mandatory in retinitis pigmentosa patients to detect exudative and telangiectatic changes at an early stage. Awareness of this potentially treatable complication of RP with a coats-like response may facilitate earlier diagnosis of the problem, leading to more timely treatment and a better prognosis.

Patient's Consent: Researchers followed the guidelines set forth in the Declaration of Helsinki.

Conflict of Interest: Authors declared no conflict of interest.

REFERENCES

1. **Pruett RC.** Retinitis pigmentosa: clinical observations and correlations. Trans Am Ophthalmol Soc. 1983;**81:**693-735.
   PMID: 6676982; PMCID: PMC1312466.
2. **Nebbioso M, Livani ML, Vingolo EM, Belcaro G, Pecorella I.** Coats'-type changes in a retinitis pigmentosa family. Panminerva Medica. 2014;**56(Suppl. 2):**15-18.
3. **den Hollander AI, Heckenlively JR, van den Born LI, de Kok YJ, van der Velde-Visser SD, Kellner U, et al.** Leber congenital amaurosis and retinitis pigmentosa with Coats-like exudative vasculopathy are associated with mutations in the crumbs homologue 1 (CRB1) gene. Am J Hum Genet. 2001;**69(1):**198-203.
   Doi: 10.1086/321263.
4. **Khan JA, Ide CH, Strickland MP.** Coats'-type retinitis pigmentosa. Surv Ophthalmol. 1988;**32(5):**317-332.
5. **Magliyah M, Alshamrani AA, Schatz P, Taskintuna I, Alzahrani Y, Nowilaty SR.** Clinical spectrum, genetic associations and management outcomes of Coats-like exudative retinal vasculopathy in autosomal recessive retinitis pigmentosa. Ophthalmic Genet. 2021;**42(2):**178-185.
   Doi: 10.1080/13816810.2020.1867754.
6. **Shields CL, Shields JA, Barrett J, De Potter P.** Vaso proliferative tumors of the ocular fundus. Classification and clinical manifestations in 103 patients. Arch Ophthalmol. 1995;**113(5):**615-623.
   Doi: 10.1001/archopht.1995.01100050083035.
7. **Hiscott P, Mudhar H.** Is Vaso proliferative tumour (reactive retinal glioangiosis) part of the spectrum of proliferative vitreoretinopathy? Eye (Lond). 2009;**23(9):**1851-1858. Doi: 10.1038/eye.2008.351.
8. **Moinuddin O, Sathrasala S, Jayasundera KT, Branham KH, Chang EY, Qian CX, et al.** Coats-like Exudative Vitreoretinopathy in Retinitis Pigmentosa: Ocular Manifestations and Treatment Outcomes. Ophthalmol Retina. 2021;**5(1):**86-96.
   Doi: 10.1016/j.oret.2020.03.026.

Authors Designation and Contribution

Sivaranjani; Consultant: *Concepts, Design, Literature search, Data acquisition, Data analysis, Statistical analysis, Manuscript preparation, Manuscript editing, Manuscript review.*

Deeksha Singh; Vitreoretina Fellow: *Design, Literature search, Data acquisition, Data analysis, Statistical analysis, Manuscript preparation, Manuscript editing, Manuscript review.*

Arun Jayanth; Vitreoretina Fellow: *Design, Literature search, Data acquisition, Data analysis, Statistical analysis, Manuscript preparation, Manuscript editing, Manuscript review.*

Manavi D Sindal; Head of Department: *Design, Literature search, Data acquisition, Data analysis, Statistical analysis, Manuscript preparation, Manuscript editing, Manuscript review.*