International Journal of Retina (*IJRETINA*) 2024, Volume 7, Number 2
P-ISSN. 2614-8684, E-ISSN.2614-8536

BILATERAL RHEGMATOGENOUS RETINAL DETACHMENT IN A YOUNG ADULT WITH NEUROFIBROMATOSIS 1 A CASE REPORT

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ABSTRACT

Introduction: *Neurofibromatosis (NF) is a set of phakomatoses involving genetic disorders, commonly associated with nerve sheath tumor development. NF1 affects various bodily systems, with ocular signs like optic pathway gliomas and Lisch nodules.*

Case report: *We present a unique case of a 33-year-old man with a classic presentation of neurofibromatosis type 1 (NF1), who sought a routine ocular examination without any specific complaints. The detailed ocular examination revealed oculus uterque (OU) retinal detachments (RD) with superonasal retinal cysts and inferior retinal dialysis in the ocular RD in NF1 patients, distinct from the previously documented cases of unilateral RD with ora serrata dialysis in NF1 patients.*

Discussion: *This case report contributes to the expanding body of literature on atypical ophthalmic presentations in NF1 patients and suggests a potential link between suboptimal fibroblastic function in NF1 and the development of retinal complications, proposing a mechanism involving faulty collagen production and subsequent vitreous base avulsion leading to retinal dialysis and detachment.*

Conclusion: *This case underscores the significance of thorough routine ocular examination in NF1 patients, emphasizing the need for a heightened suspicion of unusual ocular manifestations.*

Keywords: Retinal detachment, Neurofibromatosis type 1, Retinal dialysis, ocular manifestations
Cite This Article: TANVEER, Shafiq et al. BILATERAL RHEGMATOGENOUS RETINAL DETACHMENT IN A YOUNG ADULT WITH NEUROFIBROMATOSIS 1. International Journal of Retina, [S.l.], v. 7, n. 2, sep. 2024. ISSN 2614-8536. Available at: <https://www.ijretina.com/index.php/ijretina/article/view/284>. Date accessed: 22 sep. 2024. doi: https://doi.org/10.35479/ijretina.2024.vol007.iss002.284..

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INTRODUCTION

Neurofibromatosis (NF) comprises a group of phakomatoses characterized by various genetic disorders, often marked by the development of nerve sheath tumors. NF consist of three primary types: NF1 (prevalence of 96%), NF2 (3%), and schwannomatosis (<1%).[1] Globally, NF1 is a prevalent genetic disorder inherited as an autosomal dominant condition exhibiting age-dependent phenotypic expression.[2] NF1 impacts multiple bodily systems, leading to tumors in the central and peripheral nervous systems, including the brain, spinal cord, skin, and bones.[3]

Tumors associated with NF1 include retinal astrocytic hamartomas, multiple retinal capillary hemangiomas, and combined hamartomas involving the retina and retinal pigment epithelium. These occurrences may result in complications such as neovascular glaucoma, vitreous hemorrhage, and RD.[4] The most common tumor linked to NF1 is the neurofibroma.[5] NF1 manifests with cutaneous features like cafe-au-lait spots (most prevalent), axillary and groin freckling (referred to as Crowe sign), and dermal tumors. Ocular manifestations include optic pathway gliomas and Lisch nodules.[6] This case report details a 33-year-old male presenting with an uncommon ocular manifestation of bilateral rhegmatogenous retinal detachments (RRD) with no history of trauma or other risk factors, along with retinal cysts and dialysis at the ora serrata in the OS.

CASE REPORT

We present the case of a 33-year-old male who visited our hospital's medical outpatient department for high blood pressure. The patient was diagnosed as a classic case of NF1 based on the presence of café au lait spots, multiple subcutaneous nodules (Figure *1B*), and a positive family history of NF1. To rule out ocular manifestations of NF1, the patient underwent an ocular examination in the ophthalmology outpatient department. The patient was a known hypertensive and was on oral antihypertensives, had no reported symptoms, and his best corrected visual acuity (BCVA) was 20/200 in the OU. Autorefractor (AR) readings were -3.00 in OD and -2.00 in OS. Pupils were reactive to light, round and regular with no relative afferent pupillary defect (RAPD). Despite reporting no ocular symptoms, a routine checkup revealed Lisch nodules on the iris (Figure *1A*). The patient was phakic in the OU and had bilateral RRD (Figures *2A*, *2B*).

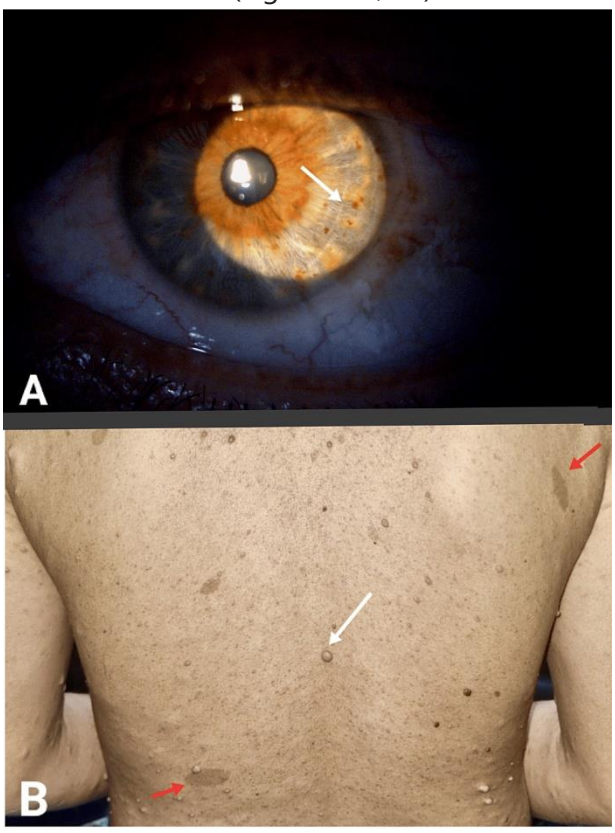

Figure 1: Classic signs of Neurofibromatosis in a 33-year-old male. The image (1A) is a slit lamp photograph (anterior segment, direct illumination) showing multiple Lisch nodules on the iris (white arrow) and (1B) shows multiple subcutaneous nodules (white arrow) and cafè au lait spots (red arrows)

In the right eye (OD) (Figure *2A*), there was a superior RRD involving the macula (macula off) with proliferative vitreoretinopathy (PVR) and a supero-temporal tear. The OS exhibited long standing RRD with PVR and superonasal retinal cysts (Figure *2B*). There was no history of contuse ocular trauma, ocular surgeries, ocular medications, diabetes, uveitis, or a family history of RRD. Clinical signs of ocular and head trauma or pathological myopia were not observed.

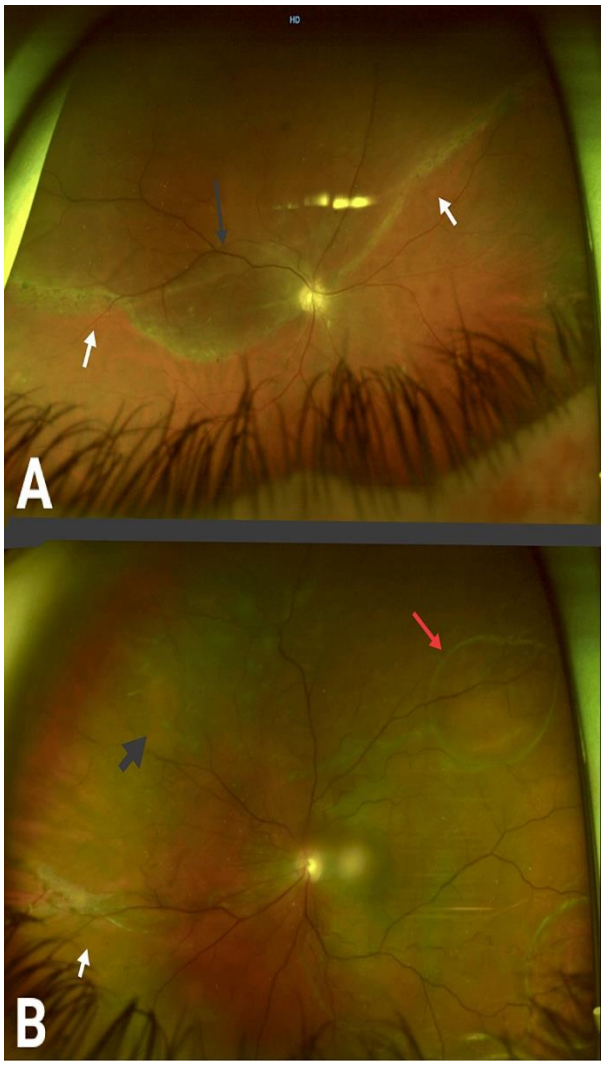

Figure 2: Colour fundus photographs showing bilateral retinal detachment in a patient with Neurofibromatosis. The image shows fundus photograph of right eye (2A) with superior retinal detachment involving the macula (white arrows) with proliferative vitreoretinopathy (black arrow). The fundus photograph of the left eye (2B) exhibits Retinal detachment (white arrow) with proliferative vitreoretinopathy (black arrow) and superonasal retinal cysts (red arrow).

Upon repeated questioning, the patient again denied any history of trauma. The exploration yielded no clinical evidence suggestive of trauma. The recommended treatment plan included a 23-G pars plana vitrectomy (PPV) with silicon oil in the OD, followed by a 360° band and 23-G PPV with silicon oil in the OS.

DISCUSSION

In 93% of adults diagnosed with NF1, the presence of Lisch nodules is reported.[7] While ocular involvement is not uncommon, the occurrence of spontaneous retinal dialysis without a history of trauma is a rare clinical phenomenon. Retinal dialysis, characterized by a circumferential tear along the ora serrata attachment, holds the potential to result in RD. Despite being typically associated with significant blunt trauma, such as sporting injuries or assaults, spontaneous retinal dialyses can also manifest, particularly in patients with hereditary factors. In their case study, Kilgore, Sanders, and Uwaydat discuss a patient with NF1 who suffered RD due to spontaneous peripheral retinal dialysis. Treatment options, including scleral buckle and PPV, were contemplated in their case.[8]

Shah et al. has documented a retrospective study based the chart reviews, which uncovered four NF1 patients who developed retinal dialysis and subsequent RRD without a history of trauma.[9] Shrestha et al. has also reported a case of a 36-year-old man diagnosed with NF1 who exhibited spontaneous ora serrata dialysis without any history of prior ocular trauma. Their case report emphasises that RD or ora serrata dialysis in NF1 may be linked to abnormal cell-matrix formation.[10]

Clemente et al., in their case report, described that patients with NF1 exhibit abnormalities in neural and neuroglial cells, along with fibroblasts. The fibroblastic cells, identified in the vitreous base

cortex, play a crucial role in collagen fibril formation. These fibrils insert through the basal lamina of the pars plana and peripheral retina, facilitating the adherence of the vitreous base to underlying tissues. In patients with NF1, suboptimal fibroblastic function may lead to faulty collagen production, resulting in deficient collagen fibers anchoring the vitreous base to the pars plana and peripheral neurosensory retina. This suboptimal function may cause spontaneous avulsion of the vitreous base, which is associated with ora serrata dialysis.[11] The avulsion of the vitreous base is often linked to retinal dialysis, tears, or vitreous haemorrhage, significantly heightening the risk of subsequent RD.[12] In a distinctive case, Jordan et al. presented a female patient with NF1, describing bilateral spontaneous vitreous base detachment without a history of ocular injury.[13]

In the comparison of PPV with and without supplemental Scleral Buckle (SB) for RRD surgery, primary reattachment rates were found to be similar. PPV exhibited lower incidences of choroidal detachment, hypotony, and strabismus/diplopia but higher rates of iatrogenic breaks and cataract development/progression than SB. Visual acuity outcomes were comparable. Another study revealed that PPV with SB had a higher single-operation success rate (SOSR) than standalone PPV, with final reattachment rates being similar. Regarding tamponade agents in RD with PVR, both perfluoropropane and standard silicone oil were considered reasonable, showing no major differences. The choice of tamponade should be individualized to each patient.[14,15]

This case report highlights an intresting incidental finding in a patient with NF1. There have been a few studies on the presence of unilateral RD in NF1 patients. However, to our knowledge, our study is the only one to demonstrate the presence of bilateral RRD in the patients with NF1.

The limitation of our study is that it is based on a single case, and while it provides valuable insights, generalising findings to the broader NF1 population may be limited. This limitation highlight the need for further research with larger sample sizes.

CONCLUSION

This case report highlights the rare association of bilateral spontaneous RDs in patients with NF1 and underscores the diverse nature of NF1 associated ocular pathology. The absence of a history of ocular trauma emphasizes the insidious nature of retinal complications in NF1 patients, urging clinicians to maintain a high index of suspicion during routine examinations. Moreover, the case emphasizes on the importance of vigilant ophthalmic examinations for early detection of ocular manifestations, even in asymptomatic patients for preserving ocular health and minimising morbidity.

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