Case Report

Combined Retinal Detachment in Von – Hipple Lindau Disease: A Case Report

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A B S T R A C T

Introduction: Retinal capillary hemangiomas (RCH) highlights the necessity of regular screening in family members patients with suspected Von-Hipple Lindau disease and importance of timely management, even in cases of associated complications.
Case report: A case of 17 years old female presenting with combined retinal detachment, secondary to retinal capillary haemangioma with strong family history suggestive of Von-Hipple Lindau (VHL) Syndrome. Pars plana vitrectomy with encircling band with trans scleral cryotherapy to hemangioma and silicone oil insertion was done to manage the case.
Conclusion: Timely screening, diagnosis and management of disease helps in preventing the morbidity and mortality associated with VHL disease

Keywords: Combined Retinal Detachment in Von, Hipple Lindau Disease

INTRODUCTION

Von –Hippel –Lindau disease (VHL) is a multisystem syndrome presenting as a benign or malignant tumor of central nervous system, pheochromocytoma, pancreatic tumors, renal cell carcinoma, retinal hemangioblastomas.[1-3] Retinal capillary hemangiomas (RCH) are often the earliest and most common manifestation[1] and occurs in 43-67% of VHL patients.[4-6] RCH is usually a solitary tumor in temporal peripheral retina, supplied by a pair of dilated and tortuous feeder vessels, but, can present as multiple retinal hemangiomas in 1/3rd of the patients. 11-15% cases can present as juxta-papillary hemangiomas. Small lesions may remain stable for years but mostly enlarge progressively and in late stages leading to combined retinal detachment, uveitis, glaucoma and pthisis.[7]

CASE REPORT

A 17 year old female presented with complaints of sudden diminution of vision in right eye since 7 days. On examination, her vision was hand movements in right eye with normal anterior segment. Fundus revealed combined retinal detachment secondary to capillary hemangioma in temporal quadrant (Figure 1a and 1b). Left eye examination was essentially normal. On enquiring, patient revealed family history of some CNS tumor in grandfather and Glomus jugulare (under treatment) in father. Clinical findings and family history pointed towards diagnosis of VHL. Patient was undertaken for Right eye Vitrectomy + belt buckle + membrane peel + endolaser + trans-scleral cryotherapy + silicone oil insertion after ruling out other systemic involvement. On follow up after 4 months her BCVA was 6/24, with attached retina under oil (Figure 2a and 2b).

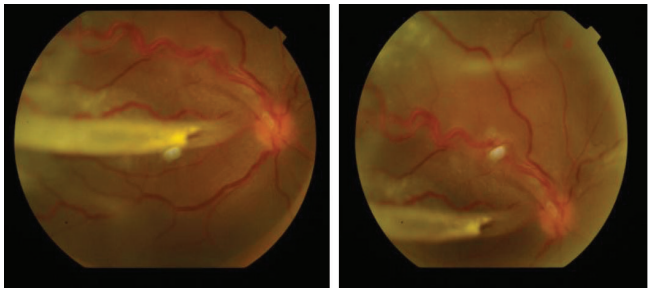

Figure-1A and B:

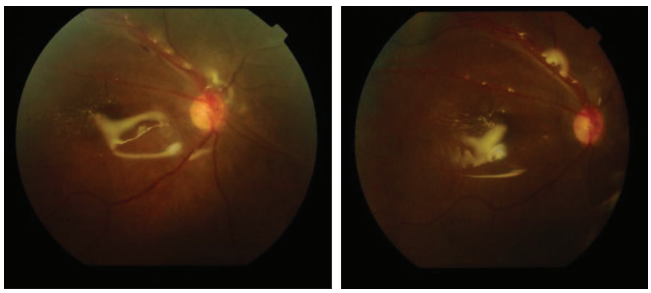

Figure-2A and B:

DISCUSSION

VHL is an inherited disease, whose gene is located on chromosome 3p and functions as a tumor suppressor gene, which spreads in an autosomal dominant pattern. It

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may have incomplete or near complete penetrance, thus importance of genetic screening in early diagnosis and hence treatment.[8] Retinal capillary hemangiomas usually present between 10-40 years of age[9] with mean age of 25 years. Few reports has indicated prevalence rate of hemangioma usually stabiloises by 30 years of age, therefore very less likelihood of developing haemangioms later on.[10] Retinal capillary hemangioma shows variable course which may vary from progression to stability to spontaneous regression. Therefore, treatment options vary from observation of small nasal or juxta-papillary tumors, while laser therapy is instituted for posteriorly located tumors and cryotherapy (double-freeze thaw technique)[9-11] for anterior ones. In case of complications like combined retinal detachment, vitrectomy is advocated and other complications like uveitis and glaucoma are managed accordingly.

Diagnosis of VHL is based on three elements: Retinal capillary hemangiomas or CNS hemangioma, visceral lesions and family history.[9-11] Once diagnosis is made, screening of patient and relatives should be done according to screening protocols, which include catecholamines levels, indirect ophthalmoscopy, CT/MRI scan.

CONCLUSION

Timely screening and hence, early management of high risk population, will help in preventing morbities and better quality of life.

Limitation

Genetic testing was not done.

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