Case Report

Von hipple- landau syndrome

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Abstract

Introduction: Von Hippel-Lindau syndrome is a multi organ disorder characterized by retinal capillary hemangiomas, CNS hemangioblastoma, various solid and cystic visceral hamartomas and malignant neoplasms. We report an 18-year-old boy who presented with unilateral acute painless loss of vision, low backache, bladder disturbancesand paresthesias in upper and lower limbs. Ocular and systemic evaluation revealed vitreous hemorrhage, cysts in kidney and pancreas and D11-12 intramedullary hemangioblastoma. Timely intervention was done to avoid the further visual loss and life threatening complications.

Keywords: Hemangioblastoma, Phakomatoses, Triple Freeze Thaw, Vitreous Hemorrhage, VonHippel- Lindau.

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INTRODUCTION

Von Hipple- Lindau syndrome (VHLS) is a part of Phakomatoses, which are group of multi system syndromes that have characteristic ophthalmic manifestations. One hundred years ago, Eugen von Hippel (1867–1938), a German ophthalmologist, published descriptions of retinal hemangioblastomas transmitted through several generations of family members. In 1926, the Swedish pathologist ArvidLindau established the link between retinal and cerebellar hemangioblastomas as well as the findings of cysts in the kidney, pancreas, and epididymis as part of a familial syndrome.

Extraocular clinical findings of VHLS

VHLS is an autosomal dominant neoplastic disorder in which multiple benign or malignant tumors and cysts with specific histopathologic features develop in the tissues of the central nervous system, including brain, spinal cord, inner ear, and retina, and in visceral organs, including kidney, adrenal gland, pancreas, epididymis, and broad ligament.

The ocular Manifestations

Consists retinal capillary hemangioblastomas, having a clinical appearance that is both characteristic and diagnostic of VHLS. These retinal lesions can be asymptomatic for years and may even regress spontaneously, but usually they grow and cause visual impairment. One of the retinal hemangioblastomacomplications is exudative maculopathy. In this report, we present a case of unilateral decrease of vision due to retinal serous detachment at the macula and vitreous hemorrhage as the initial manifestation of VHLS.

CASE REPORT

An 18-year-old boy presented with right eye sudden painless loss of vision for five days. Associated with history of low backache, bladder disturbances in form of excessive straining during micturition and alsoparenthesia in both upper and lower limbs since two months. On examination, visual acuity right eye 6/36, left eye 6/9 was recorded. Anterior segment was normal in both eyes. Intraocular pressure 14 mmHg on applanation tonometer. Fund us of right eye revealed a pair of dilated and tortuous feeder vessels coursing on the surface of the retina from the optic nerve head toward a well-defined orange-red oval shaped vascular mass in inferotemporal

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periphery (angioma). Moderate vitreous hemorrhage was seen. No abnormality was found in left eye. FFA of right eyeconfirmed clinical findings as early hyperfluorescence and late leakage of the lesion with dilated and tortuous inferotemporal feeder vessels from the disc up to the periphery. FAZ appeared to be well maintained with RP alteration inferiorly. Left eye was normal.

Figure 1: B-scan of right eye showed moderate vitreous hemorrhage with attached retina. Findings were normal in left eye

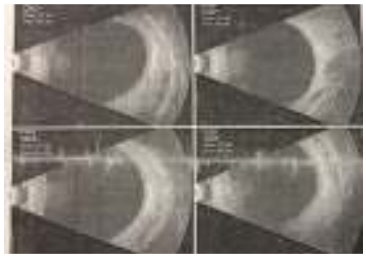

Figure 2: OCT of right eye showed subfoveal fluid with serous detachment. Left eye was normal

Figure 3: Ultrasound abdomen revealed multiple variable sized cysts in the head and body of pancreas, three variable sized cysts noted in right kidney while left kidney found normal

On MRI spine, intramedullary lesion at level of D11-12 was seen. Multiple small peritumoral cysts were associated with this lesion. Holocord syrinx extending from lumbar to cervicomedullary junction was noted. Blood and urine investigations were normal. Our patient was given triple freeze thaw technique cryotherapy to treat angioma after vitreous hemorrhage got resolved. Vision improved to 6/9 in right eye. Steroid eye drops were prescribed in tapering dose for four weeks along with anti inflammatory and mydriatic eye drops. Subsequently was followed up monthly for six months, and later on yearly. Patient was referred to other specialities and he received relevant management like D 11-12 laminectomy and excision biopsy, which revealed tumor with features consistent of hemangioblastoma.
None of his family members had similar findings.

DISCUSSION

Von Hippel-Lindau syndrome is a rare genetic disorder characterized by visceral cysts and benign tumors in multiple organ systems that have subsequent potential for malignant changes. The incidence of this disorder is approximately 1 in 36000 live births and it is inherited in a high penetrance autosomal dominant pattern. Clinical hallmarks of Von Hippel-Lindau disease are the development of retinal and central nervous system (CNS) hemangioblastomas (blood vessel tumors), pheochromocytoma, multiple cysts in the pancreas and kidneys, and an increased risk for malignant transformation of renal cysts into carcinoma. Retinal capillary hemangiomas, which are diagnosed in 50% of von Hippel-Lindau disease patients, are composed of endothelial cells and pericytes. Retinal capillary hemangiomas, usually supplied by large dilated feeder vessels, may occur in any part of the retina. Serum leakage from these vessels and hemangiomas leads to

retinal exudates. Organized fibroglial bands with traction retinal detachment and vitreous hemorrhage may occur, along with their own secondary sequelae. Tumors involving other organs and the CNS (brain, spinal cord) are present in 25% of patients with von Hippel-Lindau disease. In addition, cysts in the pancreas and kidneys may coexist.

Diagnostic Considerations

he unexpected finding of a retinal or CNS hemangioblastoma or the diagnosis of a pheochromocytoma should prompt a search for other von Hippel-Lindau disease–associated stigmata, because many of these patients meet the diagnostic criteria. Identification is important because of the increased risk of serious complications (eg, renal cell carcinoma) that are readily treated with early intervention.

Because Von Hippel-Lindau disease is a multiorgan disease that widely varies in clinical presentation, various manifestations may lead to diagnosis.

Criteria are the Following

- More than 1 hemangioblastoma in the CNS (brain, spinal cord) or eye
- A single hemangioblastoma in the CNS or retina, plus a visceral manifestation (multiple renal, pancreatic, or hepatic cysts; pheochromocytoma; renal cancer)
- Positive family history plus any 1 of the above manifestations

Management

Von Hippel-Lindau (VHL) disease is usually a progressive disorder, and ophthalmic care should begin as soon as ocular pathology is diagnosed. Surgical treatment may consist of the following:

- Argon laser photocoagulation
- Cryotherapy
- Fluid drainage
- Scleral buckling
- Vitreous surgery
- Penetrating diathermy and endodiathermy
- Radiation

In our patient cryotherapy was done which is a repetitive freeze/thaw technique used to treat anterior and larger posterior angiomas. To minimize the risk of hemorrhage, no more than 2 or 3 freeze/thaw cycles should be used per session. Multiple sessions generally are needed to arrest tumors. Resolution of the macula edema and improved visual acuity are common results of tumor eradication by cryotherapy.

CONCLUSION

The wide age range and pleiotropic manner in which Von Hippel-Lindau disease presents make the diagnosis and treatment for affected individuals, as well as their at-risk relatives, a challenge. Hence a thorough history taking including family history, ocular and systemic examination is to be emphasized. Timely diagnosis and intervention can save the patient and his family from blindness and death. The above said case conveys the importance of multidisciplinary approach and management

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