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Retinoblastoma in Adult –A Case Report and Review

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Abstract: Retinoblastoma usually presents in children younger than 5 years. To our knowledge, 22 cases of retinoblastoma in adults (older than 20 years) have been reported in the literature. Out of 28 patients with histopathologically proven retinoblastoma seen in our institute, one was adult. Patients had endophytic tumor with vitreous seeds. Ultrasonography did not reveal calcification. Immunohistochemistry with neuron-specific enolase was used to confirm the diagnosis in this case. Patient ultimately required enucleation. The diagnosis of retinoblastoma should be considered in cases of whitish mass lesion in the fundus of an adult.

Key Words: Adult Retinoblastoma; Ultra sonography; Immuno histo chemistry

I. INTRODUCTION

Retinoblastoma (RB) is a malignant neoplasm derived from photoreceptor precursor cells.[1] In more than 90% of cases, the diagnosis is established before the patient reaches 5 years of age.[2] RB is the most common intraocular malignant disease of child-hood.[3] It is rare in adults and for this reason is not usually considered in the differential diagnosis of a retinal or intraocular mass in an adult.[4]

We report a unilateral RB, with histopathological and immuno-histochemical confirmation, in a 45 year-old woman. The base of the tumor had an area of well-differentiated cells resembling retinocytoma (RC).

II. CASE DESCRIPTION

A 45 years old female was seen at our center with complaint of loss of vision in OS since 24 months. Visual acuity was 6/6 and Perception of light in OD and OS respectively. Anterior segment examination revealed no abnormality. Fundus examination of left eye showed clear media with retinal detachment having shifting fluid and small mass lesion in lower temporal area.(Figure-1) Irregular mottling was seen on the surface of mass lesion vitreous seeding was not seen. A clinical diagnosis of intra ocular tumor was made.

The right eye was normal. Ultrasonographic study and computed tomography revealed a large (8 by 12 mm), solid, noncalcified lesion.(Figure-2)

Computed tomography (CT) showed no evidence of calcification. Enucleation was performed. Grossly, the tumor

measured 12 by 8 mm. Histopathological examination confirmed the diagnosis of retinoblastoma, with few Homer-Wright rosettes and occasional fleurettes from the retina nodular tumor mass is seen.(Figure-3) on Hematoxylin and Eosin stain using 10x magnification. The mass is composed of necrotic areas. Tumor cells arranged around the blood vessels. There are dense collections of tumor cells are seen around the blood vessels. There are few Homer wright rosettes .The retinal tissue shows gliotic changes and multiple micro cystic areas are seen. Tumor cells are seen in the vitreous

There is focal choroidal invasion measuring less than 3mm.There is no optic nerve invasion. Further sections from the globe shows retina, subretinal fluid, multiple nodules of tumor cells are seen arising from ganglion cell layers of retina. Immunohistochemical study with neuron- specific enolase was positive. On last follow-up examination in, the socket was healthy, without any recurrence in the orbit.

III. DISCUSSION

RB is the most common intraocular malignant tumor of childhood and the most common tumor of the retina in the general population. [1–4] It is worldwide in distribution, affecting all racial groups.

Most cases are diagnosed before the patient is 5 years old. [1–4] The tumor is extremely rare in adults.[1–6] A literature review showed 22 histologically confirmed RBs in patients aged 20 years or older (Table 1). The median age at diagnosis was 38 (range 20–74) years. There was no predominance by either sex. The duration of symptoms before diagnosis ranged from 1 to 60 (median 16) months. Tumor location varied; in 3 cases the mass filled the eye. [5]

In adults the clinical differential diagnosis of a whitish mass in the ocular fundus includes lymphoma,

In adults the clinical differential diagnosis of a whitish mass in the ocular fundus includes lymphoma, pan ophthalmitis and inflammatory diseases of the retina. RB is usually not considered initially, because of less frequency. If inflammation, vitreous hemorrhage or cataract is present, RB diagnosis is even more difficult. [5, 13]

Imaging studies may or may not show calcifications, a characteristic of RB in children. [5]In fact, calcification is not an important finding in RB in adults. Of 16 previously reported cases, just 2 showed areas of calcification; [4–6, 9, 11] the other reports did not discuss this feature. In our patient there was no calcification.

Histopathologically, RBs are essentially undifferentiated malignant neuroblastic neoplasms that may arise in any nucleated retinal layer.[3] Characteristically, sleeves of viable cells are present uniformly along blood vessels surrounded by ischemic coagulative necrosis.[14,15] Rosette formation is highly characteristic.1–3 Fleurettes are neoplastic cells displaying photoreceptor differentiation in bouquet-like clusters.[3] In our case, typical rosettes were observed in multiple foci and were very useful in establishing a definitive diagnosis. In 16 of the 22 previously reported cases, [4–6, 9, and 11] rosettes were present: in 7 cases they were Flexner–Wintersteiner, in 4 they were Homer–Wright, and in 5 they were not specified. No areas of photoreceptor differentiation were described in the previous reports.

There is no specific immunomarker for RB. A lack of staining with HMB-45 and keratin almost rules out malignant melanoma and metastatic carcinoma respectively. As in our case, the neoplastic cells often stain positively with neuron-specific enolase. [3–6]

The positive staining with glial fibrillary acidic protein and S-100 protein is observed within the reactive stroma and not in the neoplastic cells, even in more differentiated areas.

Retinocytoma is a rare benign retinal tumor with a phenotypic RB1 gene mutation and carries genetic implications similar to those of RB.[16] It is unknown whether RB in an adult occurs de novo or is preceded by RC.[5] The presence of benign-appearing areas within an RB in an adult raises the possibility that this tumor may be this could explain the long duration of signs and symptoms in the previously reported cases.[5,9,11] Singh and colleagues[17] stated that the risk for malignant trans-formation of RC into RB is 4%. Moreover, Eagle and associates [18] described an RB with RC areas in a 7-year-old child. Of the 22 previously reported cases of RB in adults, an associated RC was described in only 2. However, it is possible that a careful histopathological review would increase the frequency of detected RCs.

Shield et al reported retrospective series of 8 cases with 6 intraocular and two orbital cases.[19]

Wells et al reported association with carcinoid tumor along with lung malignancy and recurrent retinoblastoma lesion, carcinoid tumor also may cause retinoblastoma. [18, 20]

White masses may cause confusion regarding differential diagnosis masquerade syndrome is to be differentiated in case of retinoblastoma particularly more in adults. Many malignant pathologies may result in an appearance masquerading as uveitis intraocular lymphoma, leukemia, uveal melanoma, retinoblastoma, metastatic lesions, paraneoplastic syndromes and vasoproliferative tumors. [21]

TABLE 1—SUMMARY OF REPORTED CASES OF RETINOBLASTOMA IN ADULTS

AUTHOR	A/S (Y)	DUR (M)	TUMOR SIZE	TUMOR LOCATION	HP FEATURS
MAGHY[5]	20/F	12	TOTAL	TOTAL	FW
VERHOEFF[5]	48/M	6	10*15	SN	FW
MCCREA[7]	20/M	NS	NS	NS	YES
RASMUSSEN[5]	48/M	4	16*14	ANTERIOR TO EQUATOR	NO
O'DAY[5]	29/M	NS	NS	ANTERIOR TO EQUATOR	NS
RYCHENER[5]	38/M	54	6.5*7	POSTERIOR POLE	FW
ARSENI ET AL[5]	53/M	4	NS	POSTERIOR POLE	FW
MEHRA ET AL	45/M	24	NS	ANTERIOR TO EQUATOR	FW
FINLAY ET AL[8]	74/F	6	NS	NS	YES
OHARA ET AL	43/F	48	3.8*3.8	OPTIC NERVE	HW
MAKLEY[22]	52/M	60	20*25	TOTAL	FW
LASH[9]	40/F	60	NS	NS	NO
PERZ ET AL[10]	56/M	5	NS	NS	YES
KREMLICKA ET AL[11]	42/M	4	8*18	ANTERIOR TO EQUATOR	FW
BERKELEY ET AL[23]	60/M	3	TOTAL	TOTAL	NO
TAKAHASHI ET AL[24]	26/F	6	NS	SUPERIOR	HW
NERONOVA-KOTOVA[12]	46/F	24	NS	NS	YES
NORK ET AL[6]	29/F	NS	8*10	NASAL	YES
MIETZ ET AL[4]	26/F	4	12*2	ANTERIOR TO EQUATOR	HW
BISWAS ET AL[5]	32/M	24	10*8	POSTERIOR POLE	HW
BISWAS ET AL[5]	21/M	11	3*5	SUPEROTEMPORAL	NS
BISWAS ET AL(2000)[5]	25/F	1	12.5*11.5	SUPERONASAL	NO
CURRENT REPORT	45/F	24	12*8	INFROTEMPORAL	FW

NOTE: F = FEMALE, M = MALE, NS = NOT STATED,
FW = FLEXNER–WINTERSTEINER, HW = HOMER–WRIGHT.
HP= HISTOPATHOLOGY FEATURES

CONCLUSION

When intraocular tumor is found in adult retinoblastoma should be considered as one of the differential diagnosis.

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Legends:
Figure.1 Retinal image
Figure.2 Ultra sonography B scan image
Figure.3 Histopathology image

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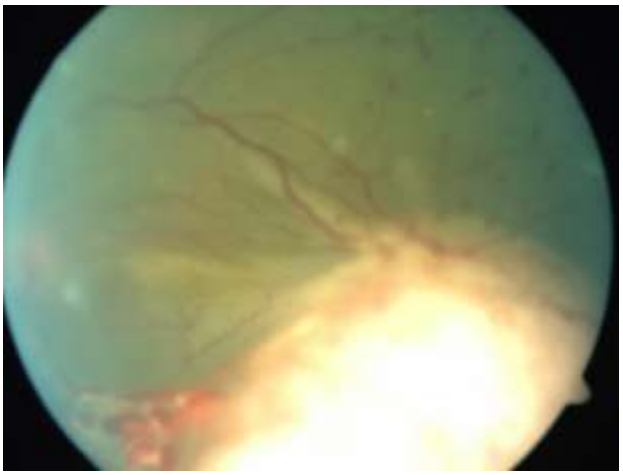

Figure-1

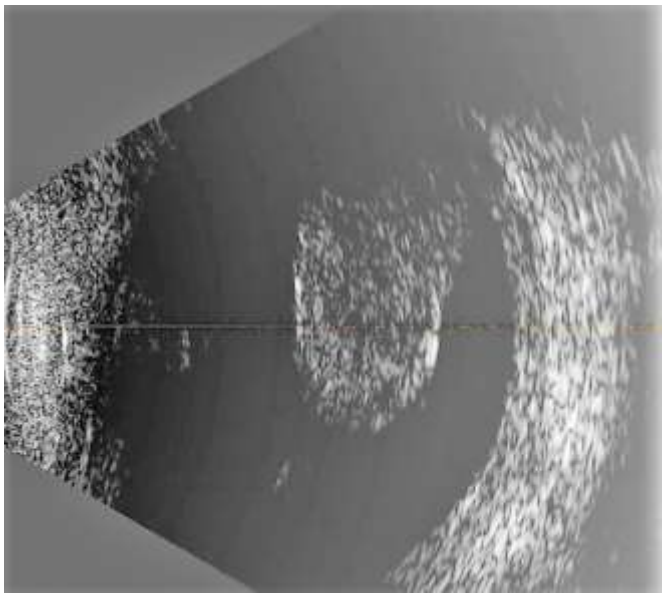

Figure-2

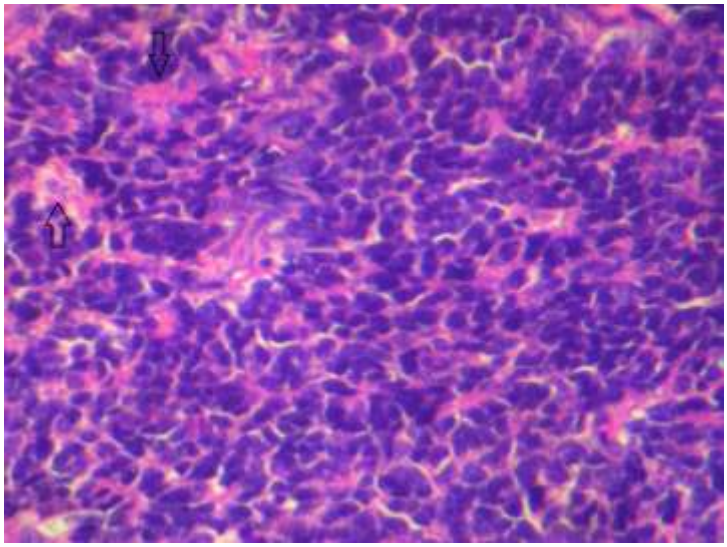

Figure-3

