Choroidal Melanoma in Association with Juxtapapillary Melanocytoma

A 68-year-old Saudi man presented with reduced vision of his left eye for duration of 4 months. On examination, his vision in the affected eye was counting fingers near the face. The left fundus (Fig 1) showed an elevated round choroidal mass, partially overlying the optic disc infero-temporally. The lesion was dark brown with jet black color peripherally with an associated sub retinal hemorrhage. Ultrasound showed a dome shaped posterior surface surrounding the optic disc, maximum elevation of the lesion was seen infero-temporally and had moderate internal reflectivity with moderate sound attenuation. Doppler ultrasound showed a vascular mass. Intraocular fluoroscein angiography was not conclusive because of poor fundus detail. Our patient presented as choroidal melanoma probably evolving from pre-existing juxtapapillary melanocytoma.

Radiological studies did not show extraocular extension, hyperechogenic or evidence of long term, however, MRI of the left orbit was suggestive of extension of the lesion into the optic nerve head.

The case was discussed and enucleation of the globe was decided (fig 2, 3). Histopathology showed a darkly pigmented juxtapapillary choroidal tumor with overlying retinal detachment and collection of sub retinal fluid (fig 4). Bleached sections showed two types of cells: 1) round to oval with abundant cytoplasm, pigment granules and small nucleus representing melanoma cells and 2) spindle-B cells representing the developing melanoma with atypical mitotic figures identified, which invade the adjacent optic nerve. This was further supported by positive immunostaining with HMB45. Immunostaining with Melan-A showed no expression (original magnification 4,000).

Few macrophages with phagosomes containing irregular melanin granules were identified (fig 6). Some balloon cells were also seen (fig 7). The tumor invaded the adjacent optic nerve head (fig 8 & 9). Diagnosis was choroidal melanoma in association with a pre-existing juxtapapillary melanocytoma. This was further supported by positive immunostaining with HMB45 (fig 10).

Based on our finding, we presume that the patient had pre-existing atypical juxtapapillary melanocytoma and the recent deterioration of the patient’s vision was caused by the rapid tumor growth, visual loss due to hemorrhage and subretinal hemorrhage. However, we do not have any documentation that the patient had a pre-existing melanocytoma lesion previously.

Discussion

Dendritic melanocytes, which are derived from the neural crest, are considered to be the origin of pigmented intracocular growths such as, nevi, melanocytoma and melanoma. Melanocytoma is a characteristic darkly pigmented tumor which can occur anywhere in the uvea where melanocytes are present, and classically in the optic nerve head1. A few cases of choroidal melanoma arising from optic nerve melanocytoma have been reported but this issue for some authors was controversial. Apple and associates2 reported a case of malignant transformation from optic nerve melanocytoma. The initial gray mass at the superior aspect of the disk was not recorded and seemed atypical in location. It was not clear if melanoma originated from the choroid or from the optic nerve. At the Verheff Ophthalmic Pathology Society meeting in 1975, Zimmerman presented a case that was initially diagnosed as low-grade melanoma arising from a melanocytoma of the optic disc, but the final pathological diagnosis was melanocytoma of the optic nerve head as reported by Mansour and associates3. Shields and associates4 reported a choroidal melanoma developing from a melanocytoma that affected both the optic disc and choroidal melanoma. The malignant tumor in this case was of the mixed-cell type.

De Potter and associates5 reported a mixed-cell malignant melanoma confined to the optic disc. It was considered a rare example of malignant transformation of a melanocytoma even without focal loss of visible melanocytes histologically. In 1992, Loeffler6 reported a similar case of an opticallyscopically typical melanocytoma of the optic disc which turned into a malignant melanoma of spindle-B cell type located within the choroidal stroma but not to the optic disc over 7 years. Tumor extension through Bruch’s membrane and in the nerve fiber layer of the optic nerve head was noted. On the other hand, no “melanocytoma cells” were seen within the choroid. They were able to identify two types of cells by ultrastructure studies: one of macrophage origin with large melanomas, and the other with elongated nuclei, many cytoplasmic organelles and smaller melanomas. The pre-melanomas showed a granular or filamentous substructure. However the two distinct types of melanoma cells described by Jarez and Tso7 couldn’t be demonstrated in his case. Meyer and his colleagues8 reported a typical melanocytoma of the optic disc with documented change in color and size eventually with dramatic visual loss 5.5 years after the initial diagnosis. The histopathology demonstrated spindle-B malignant melanoma cells interwoven with the population of the melanocytoma cells which extended to the lamina crib Rosai of the optic nerve. Therefore, they strongly recommended regular examination with serial fundus photographs of melanocytoma cases.

More recently, in a large study, Shields and associates9 reported the rate of malignant transformation of the disk melanoma 1% to 2%.

Our patient presented at a later stage with recent onset of visual loss and an elevated lesion suggestive of malignancy. Unfortunately, no previous clinical or photographic documentation of his original melanocytoma was available; however, there was enough histopathologic and ultra structural evidence of a pre-existing juxtapapillary melanocytoma within the infero-temporal choroid which is the usual location as has been observed by Zimmerman. The typical polyhedral large heavily pigmented cells were mixed with spindle-B melanoma cells. The malignant cells superficially invaded the optic nerve head. The ultrasonographic studies showed mainly type 2 melanoma cells with a markedly indented nucleus and relatively smaller melanin (fig 11). Few macrophages with phagosomes containing irregular melanin granules were identified (fig 12). Our case further supports the necessity of close follow-up of the melanocytoma cases.

In conclusion, we report one of the few well-documented cases of choroidal melanomas that was associated with juxtapapillary melanocytomas and was confirmed by histopathological study. We believe that periodic follow-up of the patient with optic disc melanocytoma is necessary.

References

8. H_EXTENSIONS