Choroidal Melanoma in a 14-Year-Old Patient With Ocular Melanocytosis

Kaan Gündüz, MD; Jerry A. Shields, MD; Carol L. Shields, MD; Ralph C. Eagle, Jr, MD

A 14-year-old male adolescent with ocular melanocytosis and secondary glaucoma in the left eye had a 2-year history of a progressively enlarging fundus lesion. Ocular examination revealed diffuse hyperpigmentation of the episclera and a smooth velvety thickening and hyperpigmentation of the left iris. Ophthalmoscopy disclosed diffuse choroidal pigmentation and a pigmented mass that occupied the macular area and surrounded the optic nerve. Ultrasonography showed an acoustically hollow lesion with scleral bowing and choroidal excavation. Based on clinical and ultrasonographic findings, the diagnosis was choroidal melanoma in a young patient with ocular melanocytosis. The eye was enucleated. Histopathologic examination revealed ocular melanocytosis with diffuse uveal melanocytosis and amelanotic malignant melanoma of the choroid. The choroidal melanoma apparently arose from a preexisting choroidal nevus. Even young patients with ocular melanocytosis should have regular follow-up examinations for early detection of uveal melanoma.

Congenital ocular melanocytosis is an abnormality characterized by diffuse brown melanocytic hyperplasia in the sclera and uvea. In congenital oculodermal melanocytosis (nevus of Ota), there is cutaneous hyperpigmentation in the distribution of the trigeminal nerve in addition to ocular involvement. Congenital ocular and oculodermal melanocytosis affect only about 0.04% of the white population. An association between ocular and oculodermal melanocytosis and the development of uveal, orbital, and meningeal melanoma is well recognized. The presence of increased numbers of melanocytes in these affected tissues may provide the basis for susceptibility to the development of melanoma.

Uveal melanoma occurs at an average age of 55 years and is rare in children. In one report, only 40 (1.1%) of 3706 consecutive patients with uveal melanoma were found to be younger than 20 years. We describe a 14-year-old patient who had ocular melanocytosis and a choroidal melanoma.

REPORT OF A CASE

A white male infant was found to have episcleral pigmentation consistent with ocular melanocytosis in the left eye at age 9 days.

From the Oncology Service (Drs Gündüz, J. Shields, and C. Shields) and Pathology Department (Dr Eagle), Wills Eye Hospital, Thomas Jefferson University, Philadelphia, Pa.
choroidal excavation, scleral bowing, and a thickness of 4.9 mm. The discrepancy between the clinical estimate and ultrasonographic measurement of the tumor thickness was probably caused by the posterior scleral bowing induced by the rapidly growing tumor. The long history of glaucoma, which caused advanced glaucomatous cupping, might have contributed to the development of posterior bowing.

After a diagnosis of choroidal melanoma was made, the patient was treated with enucleation and implantation of a hydroxyapatite implant. He has been followed up for 60 months and has no evidence of an orbital or central nervous system tumor or systemic metastases.

Findings from pathologic examination of the enucleated globe revealed diffuse uveal melanocytosis that was particularly prominent anteriorly and in the posterior choroid. The iris (Figure 2 and Figure 3) and ciliary body (Figure 3) were heavily pigmented and massively thickened by an infiltrate of nevus cells that had bland nuclear characteristics. The massive melanocytic proliferation obstructed much of the anterior chamber angle, but some parts of the trabecular meshwork remained open (Figure 3). Posteriorly, an oval focus of lightly pigmented tumor was noted in the thickened and heavily pigmented choroid (Figure 4). The tumor measured 7 × 6 × 3 mm. Microscopy disclosed that the lightly pigmented focus was a spindle cell melanoma comprised of a syncytium of spindle A and B cells with oval nuclei and distinct nucleoli (Figure 5). Two mitotic figures were counted in 40 high-power fields. Several vascular loops were present, but necrosis and lymphocytic infiltration were not observed. The cells in the nevus bordering the melanoma were plump and polyhedral with copious quantities of intensely pigmented cytoplasm, and bland nuclear characteristics were disclosed by bleach sections (Figure 6). There was marked choroidal, scleral, and episcleral pigmentation consistent with uveal melanocytosis (Figure 4).

**COMMENT**

Conditions that are associated with or predispose to the development of uveal melanoma include ocular and oculodermal melanocytosis, familial atypical mole-melanoma syndrome, Li-Fraumeni syndrome, familial melanoma, and neurofibromatosis type 1. A relationship between congenital oculodermal melanocytosis and early-onset uveal melanoma is recognized. In one series, 4 of 7 patients younger than 20 years with choroidal melanoma were found to have ocular melanocytosis. Familial atypical mole-melanoma syndrome, Li-Fraumeni syndrome, familial melanoma, and neurofibromatosis type 1 have also been associated with early-onset uveal melanoma. Neurofibromatosis type 1, on the other hand, has not been found to predispose toward the develop-
ment of uveal melanoma in children.13

In general, congenital ocular and oculodermal melanocytosis are the predisposing conditions most commonly associated with uveal melanoma. Sixty (1.3%) of 4500 consecutive patients with uveal melanoma evaluated by an ocular oncology referral center during a 17-year period had ocular or oculodermal melanocytosis.14,15 There have been several reports of bilateral and multiple choroidal melanomas that occurred in patients with ocular melanocytosis.16,17 Furthermore, several disparate melanocytic tumors such as choroidal melanoma and ciliary body melanocytoma also have been reported to develop in a single eye with ocular melanocytosis.18 Therefore, patients with ocular or oculodermal melanocytosis are prone to develop both benign and malignant ocular tumors.

It has been calculated that only 1 in every 400 patients with ocular or oculodermal melanocytosis will develop uveal melanoma during their lifetime. In 99% of these patients, the uveal melanoma occurs after age 10 years.19 The diagnosis of choroidal melanoma at age 14 years in our patient is in accordance with this estimated susceptible period of tumor development.

Choroidal melanoma can arise from a preexisting choroidal nevus.20 Findings from histopathologic examination of the enucleated eye in our patient confirmed that the choroidal melanoma arose from a distinct choroidal nevus with magnocellular features within the diffuse uveal melanocytosis. The initial pigmented fundus lesion detected at age 12 years was probably a choroidal nevus that underwent transformation into a melanoma during the next 2 years in the presence of ocular melanocytosis.

This case also demonstrates that patients with congenital ocular melanocytosis can develop glaucoma. The infiltration of the trabecular meshwork by the neural crest–derived melanocytes is an important factor in the development of glaucoma in these patients.19 Abnormal development of trabecular meshwork cells, which are also of neural crest origin, may also play a contributory role in the pathogenesis of glaucoma.20

In conclusion, patients with congenital ocular and oculodermal melanocytosis, regardless of their age, should be regularly followed up for the development of glaucoma and uveal melanoma. The risk of developing uveal melanoma increases gradually after the first decade of life.

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REFERENCES


