Surgical Resection of a Retinal Pigment Epithelial Hamartoma

Several darkly pigmented tumors affecting the macular area have been described, including retinal pigment epithelial (RPE) hamartomas,1 combined retina-RPE hamartomas, melanocytic nevi of the RPE (congenital hypertrophy of the RPE), choroidal nevus, and melanomas invading the retina. Melanocytomas can rarely also be located in the macular area without involving the overlying retina.

Retinal pigment epithelial hamartomas are focal, nodular, jet-black lesions that usually appear to involve the full thickness of the retina and to spill onto the inner retinal surface in an umbrella fashion, frequently in the macular area.1 Combined retina and RPE hamartomas can be papillary or nonpapillary, variably elevated pigmented lesions at the level of the RPE with a thin semitransparent membrane partly covering the tumor surface.1 There are various reports in the literature of vitrectomy to peel the epiretinal component of these lesions,2,3 with improvement of visual acuity. Melanocytoma is described as a darkly pigmented lesion of variable size and shape located partly or completely in the optic nerve head. Since the clinical and histopathologic nature of the lesion was clarified, similar tumors have been reported in other locations,4,5 including the macula, where they always involve the choroid only.

We report an unusual pigmented macular tumor in a child that was surgically excised. It was also studied histopathologically and diagnosed as an RPE hamartoma.

Report of a Case. A 5-year-old white boy of Mediterranean descent was referred to us because of decreased visual acuity and a central scotoma in his left eye. The patient was healthy and his medical history was unremarkable.

On examination, the patient’s best-corrected visual acuity was 20/25 OD and 20/200 OS without esotropia. Results of examination were normal except those in the fundus of the left eye. Fundus examination of the left eye showed a round, nodular-shaped, darkly pigmented mass involving the central macular area (Figure 1). The maximum diameter and thickness measured by tridimensional ultrasonography were 1.2 mm and 1.6 mm, respectively. The mass had a granular surface and very sharp margins. Dilated tortuous retinal vessels appeared to be drawn inward toward the base of the tumor. Since the clinical and histopathologic nature of the lesion was clarified, similar tumors have been reported in other locations,4,5 including the macula, where they always involve the choroid only.

Fluorescein angiography showed dilation and marked tortuosity of the retinal vessels surrounding the base of the tumor, suggesting the presence of an epiretinal fibrous membrane (Figure 2 and Figure 3). Computerized perimetry confirmed the presence of a central scotoma corresponding to the area of the tumor.

The extremely dark pigmentation was similar to that observed in melanocytomas and melanomas, but...
the unusual location, the absence of fibrillated margins, the epiretinal membrane, and the green-brown reflex in the subretinal area suggested otherwise. Because the diagnosis was not reached with clinical examination and noninvasive tests, we decided to perform a vitrectomy to obtain a sample for diagnostic biopsy, relieve the retinal traction around the tumor, and, if possible, resect the tumor completely.

A 3-port pars plana vitrectomy was performed, and the posterior hyaloid membrane was dissected. With a bent microblade, a dissection plane was created to release the traction over the retina surrounding the tumor. Between the inner retina and the outer part of the tumor, a dissection plane was found, and by means of a bimanual technique, the entire tumor was resected and removed completely from the retina, leaving a craterlike configuration in the macular area (Figure 4).

The postoperative period was uneventful. The craterlike appearance of the macular area resolved by 1 month postoperatively, and 1 month after surgery visual acuity had not changed from the previous 20/200. Treatment of the amblyopia with occlusion of the right eye achieved a final visual acuity of 20/80. The central scotoma resolved.

By light microscopy, the tumor was composed of tightly packed, maximally pigmented, round cells (Figure 5). On bleached preparations (Figure 6), the cells were large and plump, round, or polyhedral with abundant eosinophilic cytoplasm and uniform, ovoid or round, benign-appearing nuclei. The cells expressed S100 protein (Figure 7), and epithelial markers were found to be positive (Figure 8) with the use of cytokeratin markers. Mitotic figures and other evidence of anaplasia were not observed. No newly formed blood vessels were found within the tumor.6

Comment. This case proved to be an unusual diagnostic problem from both clinical and anatomic standpoints. There are 2 melanin-bearing cells of the eye that may cause pigmented tumors: uveal melanocytes and RPE cells. Uveal melanocytes may produce disorders such as melanocytoma, nevus, and melanoma. Melanocytomas are considered to be derived from a massive accumulation of ectopic uveal melanocytes during embryogenesis and classically affect a portion of the optic nerve. A melanocytoma that is located in the retina without involving the optic disc is exceptional. Recently, Jürgens et al5 observed a presumed retinal melanocytoma involving the macular area, but it was not confirmed histopathologically.

The RPE cells may produce congenital hypertrophy, hamartomas, adenomas, and adenocarcinomas. Retinal pigment epithelial hamartomas, also referred to as “congenital pigment epithelial adenomas” or “primary pigment epithelial hyperplasia,” are focal, nodular, jet-

Figure 5. Histopathologic studies by light microscopy showing that the tumor is composed of tightly packed, maximally pigmented, round cells. No cellular detail could be observed by staining with hematoxylin-eosin (A, original magnification ×250; B, original magnification ×1000).

Figure 6. A bleached preparation from hematoxylin-eosin staining showing a uniform cell population (cells 15 to 25 mm in diameter) composed of large, round cells with abundant cytoplasm and round, centrally placed nuclei with occasionally conspicuous nucleoli (original magnification ×1000).
black lesions that usually appear to involve the full thickness of the retina and often extend onto the anterior surface of the retina in a mushroom configuration.1 Recently, a study of 5 cases of hamartoma of the RPE was published by Shields et al,7 in which the authors described 5 cases similar to ours, but all of them adjacent to the fovea and with good visual acuity. 

Clinically the tumor could be classified as an RPE hamartoma because the age of the patient, the jet-black lesion involving the retina, and the mushroom shape in the macular region favored this diagnosis. The appearance of the tumor and its location were similar to an RPE hamartoma reported by Gass.1 However, the prominent shape and the margins, the mushroom appearance on B-scan ultrasonography, and the green-brown reflex in the subretinal area suggested a uveal melanoma with retinal involvement. 

We observed intraoperatively that the tumor was located over the retina, and after its dissection and removal, the full-thickness retina, with the xanthophyll pigment, and the fovea were found behind the tumor. Therefore, the classic assumption that the RPE hamartoma originated in the RPE, with posterior involvement of the full-thickness retina extending through the internal limiting membrane onto the inner retinal surface, was not confirmed in our case, although Gass1 also described hamartomas with a superficial location. In the present case, tumor excision permitted a complete histopathologic examination and made it possible to establish the diagnosis of RPE hamartoma, rule out the possibility of malignancy, and improve the visual acuity after amblyopia therapy was performed.

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