Chapter 34
Melanocytoma

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34.1 Definitions

Melanocytoma (magnocellular nevus) is a benign, congenital hamartoma arising from melanocytes.

34.2 Symptoms

The majority (86 % of cases) are asymptomatic. A small proportion of patients may experience symptoms including decreased visual acuity, floaters, and/or flashes of light.

34.3 Signs

Melanocytoma appears clinically as a dark brown to black lesion, often with feathery margins, which frequently occurs at the optic nerve head. The lesion sometimes extends into the peripapillary retina and choroid (Fig. 34.1). Melanocytoma is nearly always unilateral. Associated findings may include relative afferent pupillary defect (RAPD) in the affected eye and visual field defects including enlargement of the blind spot. Ophthalmologists should be aware of rare complications of melanocytoma such as retinal edema, retinal exudation, associated hemorrhage, and vascular occlusion.

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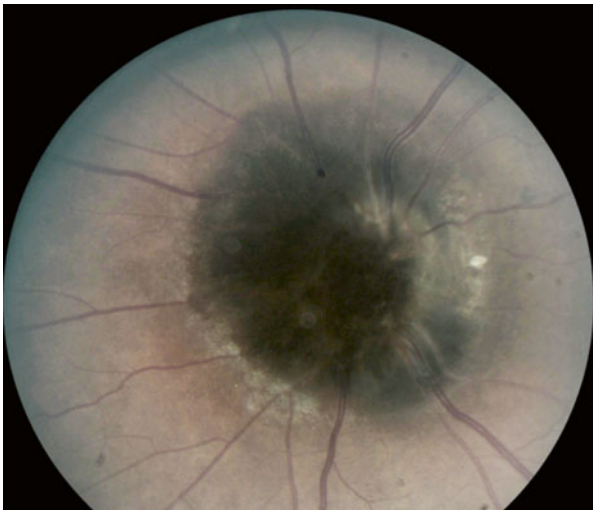

Fig. 34.1 Optic disk melanocytoma demonstrating a uniformly dark mass with feathery borders involving the surrounding retina and choroid. Deposition of calcium is observed

34.4 Epidemiology

The mean age of diagnosis is 50 years. There is a slight predilection for females (female/male ratio = 3/2) and a racial predisposition, with approximately one-third to one-half occurring in persons of African ancestry. Caucasian individuals with melanocytoma are more commonly of Hispanic or Italian descent.

34.5 Predisposing Conditions

Melanocytoma has no strong association with systemic disease and is generally considered to occur sporadically. There have been reports of optic disc melanocytoma occurring with intracranial meningioma and ocular melanocytosis.

34.6 Differential Diagnosis

Choroidal melanoma is the most important entity to consider in the differential diagnosis of melanocytoma. Other diagnoses include choroidal nevus, congenital hyperplasia of the retinal pigment epithelium (CHRPE), combined hamartoma of the retina and RPE, RPE adenoma, and juxtapapillary subretinal hemorrhage.

34.7 Etiology

These lesions occur sporadically. The etiology of optic disc melanocytoma is unknown.

34.8 Workup/Testing

The diagnosis of a melanocytoma of the optic disc is generally straightforward and can be made with dilated ophthalmoscopic examination alone. Formal visual field testing and fundus photography should be performed at the time of diagnosis to document baseline characteristics. Ancillary imaging can also be helpful:

- Optical coherence tomography (OCT): useful in detecting microscopic extension of the tumor into the retrolaminar portion of the optic nerve as well as in determining the extent of subretinal fluid and macular edema which may not be apparent with ophthalmoscopy alone [8].
- Fluorescein and indocyanine green angiography (FA and ICG): useful for differentiating melanocytoma from melanoma, as the former demonstrates dense hypofluorescence corresponding to the location of the tumor. In contrast to uveal melanoma, intrinsic vasculature is not present.
- Ultrasonography (B/A scan): reveals an acoustically solid mass (Fig. 34.2) with high internal reflectively.

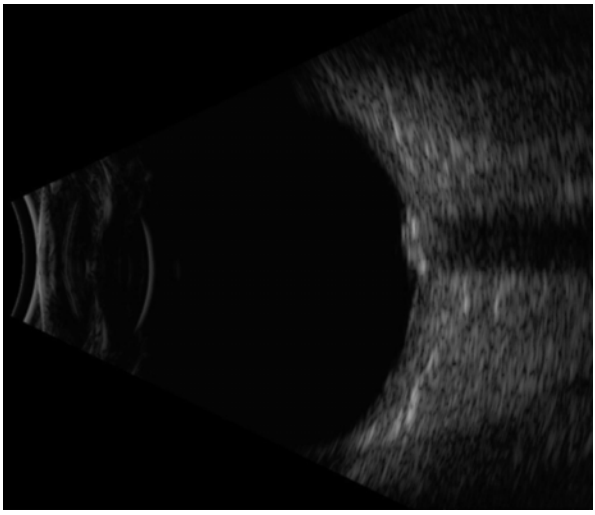

Fig. 34.2 B-scan ultrasonography reveals a solid, dome-shaped, regularly structured lesion overlying the optic disc. Maximal elevation measures 1.6 mm. There is adjacent orbital shadowing secondary to calcification

34.9 Prognosis and Management

An initial tumor thickness of ≥1.5 mm is a risk factor for growth. A small fraction, 1–2 % percent of optic nerve melanocytoma, transforms to malignant melanoma. Rapid visual loss may result from central retinal vascular occlusion, ischemic optic neuropathy associated with tumor necrosis, or malignant transformation. If the vision loss is accompanied by progressive growth, malignant transformation should be suspected and treatment should be initiated. Unfortunately, there are no current therapies to prevent growth of optic nerve melanocytoma.

34.10 Follow-Up

For typical, asymptomatic melanocytoma, annual dilated fundus examination with fundus photography and formal visual field testing is recommended. When melanocytoma demonstrates atypical features, patients should be followed at closer intervals to exclude the possibility of malignant transformation.

References and Suggested Reading

1. Zimmerman LE, Garron LK. Melanocytoma of the optic disk. Int Ophthalmol Clin. 1962;2:431–40.
2. Joffe L, Shields JA, Osher RH, et al. Clinical and follow-up studies of melanocytomas of the optic disc. Ophthalmology. 1979;86:1067–83.
3. Shields JA, Demirci H, Mashayekhi A, et al. Melanocytoma of optic disc in 115 cases: the 2004 Samuel Johnson Memorial Lecture, part 1. Ophthalmology. 2004;111:1739–46.
4. Shields JA, Demirci H, Mashayekhi A, Eagle RC, Shields CL. Melanocytoma of the optic disk: a review. Surv Ophthalmol. 2006;51:93–104.
5. Turell ME, Singh AD. Nonvascular hamartomas. In: Wright KW, Spiegel PH, editors. Pediatric ophthalmology and strabismus, vol. 1. 3rd ed. New York: Springer; 2012. p. 928–37.
6. Walsh TJ, Packer S. Bilateral melanocytoma of the optic nerve associated with intracranial meningioma. Ann Ophthalmol. 1971;3:885–8.
7. Shinoda K, Hayasaka S, Nagaki Y, et al. Melanocytoma of the left optic nerve head and right retrobulbar optic neuropathy compressed by a tuberculum sellae meningioma. Ophthalmologica. 2000;214:161–3.
8. Shields CL, Perez B, Benavides R, Materin M, Shields JA. Optical coherence tomography of optic disk melanocytoma in 15 cases. Retina. 2008;28(3):441–6.