



A 17-Year-Old Male with Optic Nerve Pit - Barton Blackorby, MD; Thomas K. Kruppenacher, MD & Kevin J. Blinder, MD



Introduction:

A 17-year-old male was referred for evaluation of abnormal optic nerve appearance of the left eye. The patient is a known migraineur. He denied flashes, metamorphopsia, or blurry vision.

Exam:

His best corrected visual acuity was 20/20 OU. Intraocular pressures were within normal limits. There was no afferent pupillary defect. Visual fields were full to confrontation and extraocular motility was full. Anterior segment exam was normal.

Dilated fundus examination revealed a normal optic nerve in the right eye, and a 0.2 cup-to-disc ratio in the both eyes. The left optic nerve was also notable for a temporal oval gray excavation consistent with an optic nerve pit, peripapillary atrophy, and a developing PVD (Figure 1). The macula examination was normal as well as the peripheral exam. OCT imaging revealed an optic nerve pit and a normal macula (Figure 2).

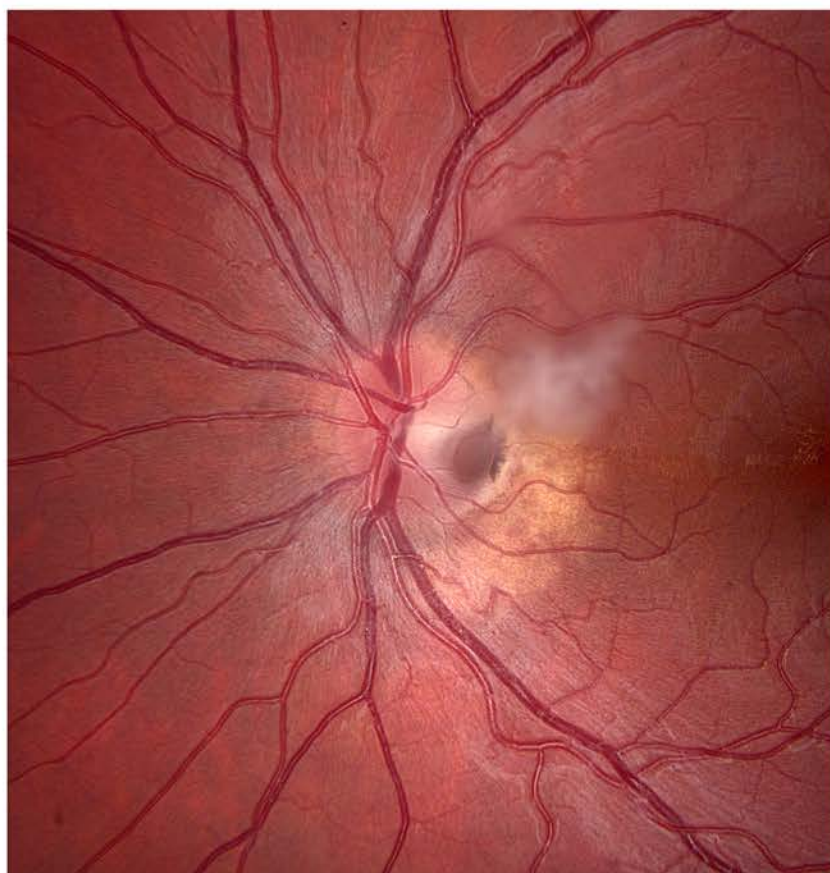


Figure 1: Color photo of left optic nerve.

Discussion:

Optic nerve pits are congenital excavations of the optic nerve head, first described by ophthalmologist Dr. Wieth in 1882.¹ They are most often unilateral, show no gender predilection, nor exhibit genetic inheritance and have a prevalence of one in 11,000 patients. Optic nerve pits are grayish in color and oval or round in shape. Although most commonly found along the temporal aspect of the optic nerve, they can occur any-

where along the optic nerve margins. Questionable diagnoses are usually confirmed via OCT which readily reveals the presence of an optic nerve pit and any associated subretinal fluid. Fluorescein angiography is typically unremarkable and does not show leakage of dye into the area of macular detachment (Figure 3).

Clinically, they are most often asymptomatic.³ Enlarged blind spots reflecting any areas of involved nerve fiber layer abnormalities are normally the only consequences of their presence, and noticed only on formal visual field testing. Optic nerve pits can become symptomatic

when they allow for the formation of subretinal fluid and localized serous macular detachments (Figure 4). Serous macular detachments generally develop in adulthood in approximately 25 to 75% eyes with optic nerve pits.⁴⁻⁷ This complication presents with blurry vision and metamorphopsia which is why questioning of these symptoms is important even on routine follow-up exam.

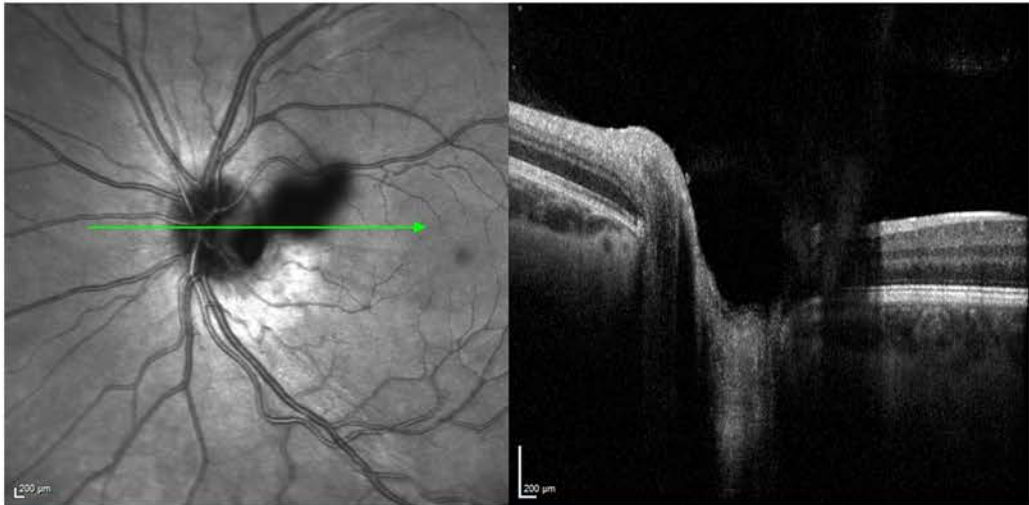
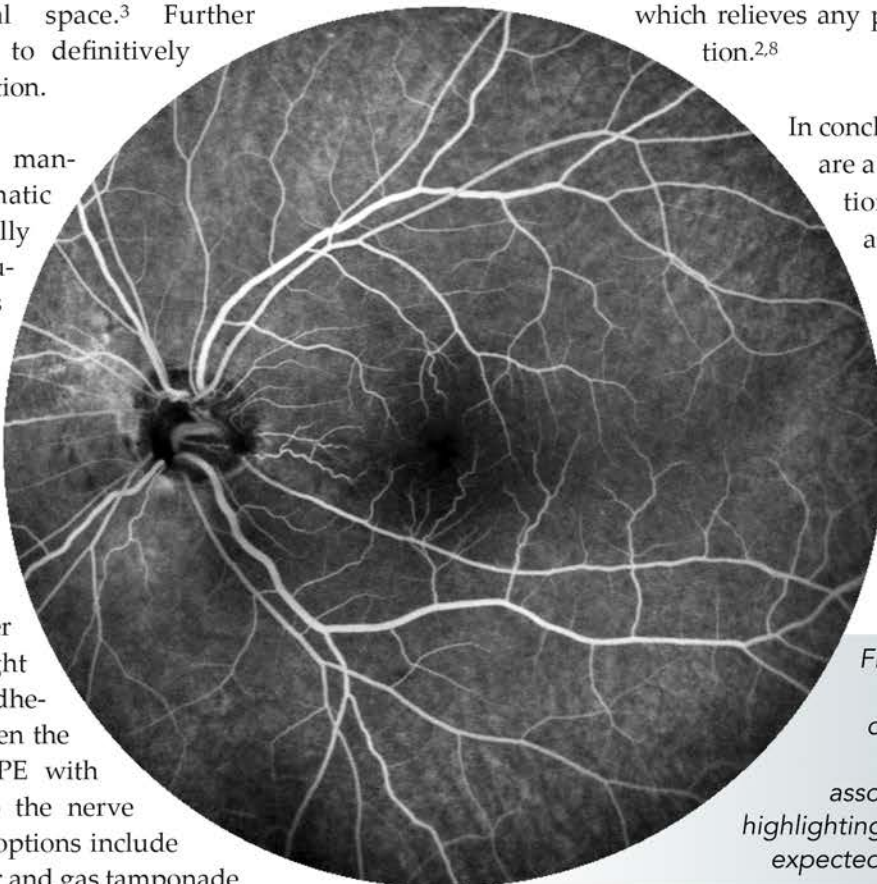


Figure 2: OCT imaging of the left optic nerve.

There are two leading hypotheses concerning the pathophysiology of optic pit associated maculopathy; fluid enters the subretinal space from the vitreous cavity or the subarachnoid space. A recent study using scanning electron microscopy to evaluate optic nerve pits and post-mortem eyes revealed that some optic nerve pits exhibited a defect in the presence of the normal diaphanous membrane overlying the optic nerve. This defect may provide access for vitreous fluid to enter into the adjacent subretinal space.³ Further research is needed to definitively evaluate this association.

Concerning clinical management, asymptomatic patients can typically be followed with routine exams. For cases complicated by persistent or recurrent sub-macular fluid, laser photocoagulation to the peripapillary retina adjacent to the pit can be performed. The goal of this laser is to achieve a light burn resulting scar adhesions forming between the outer retina and RPE with minimal damage to the nerve fiber layer. Surgical options include vitrectomy with laser and gas tamponade

which has the added benefit of both relieving any posterior vitreous traction and promoting resorption of subretinal fluid secondary to the increased surface tension of the gas. Internal limiting membrane peeling is a newer approach with multiple studies showing improvement for persistent maculopathy status post prior vitrectomy, laser, and gas.² Macular buckling has also been described in severe cases, which involves placement of a scleral buckling agent over the macula which relieves any posterior hyaloid traction.^{2,8}



In conclusion, optic nerve pits are a rare congenital condition that are typically asymptomatic in the absence of associated maculopathy. Management of these patients should be tailored to their symptoms. Home monitoring with Amsler grid is recommended.

Figure 3: Fluorescein angiography of a different patient with optic nerve pit and associated maculopathy highlighting the lack of leakage expected with this condition.

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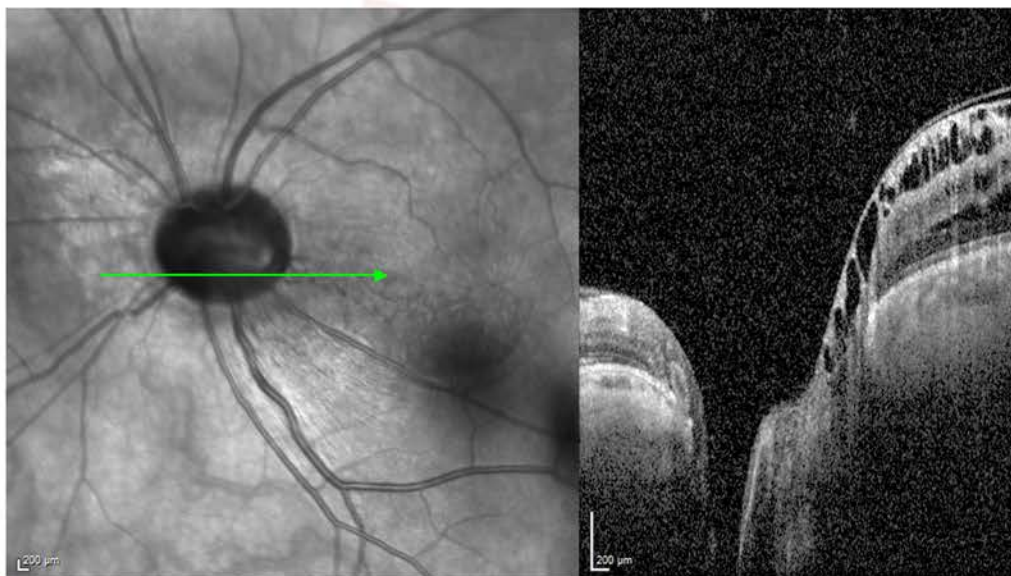


Figure 4: OCT imaging of the patient from Figure 3 showing macular schisis and edema secondary to his optic nerve pit.

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