An Asymptomatic 35-Year-Old Man Referred for Retinal and Optic Nerve Evaluation

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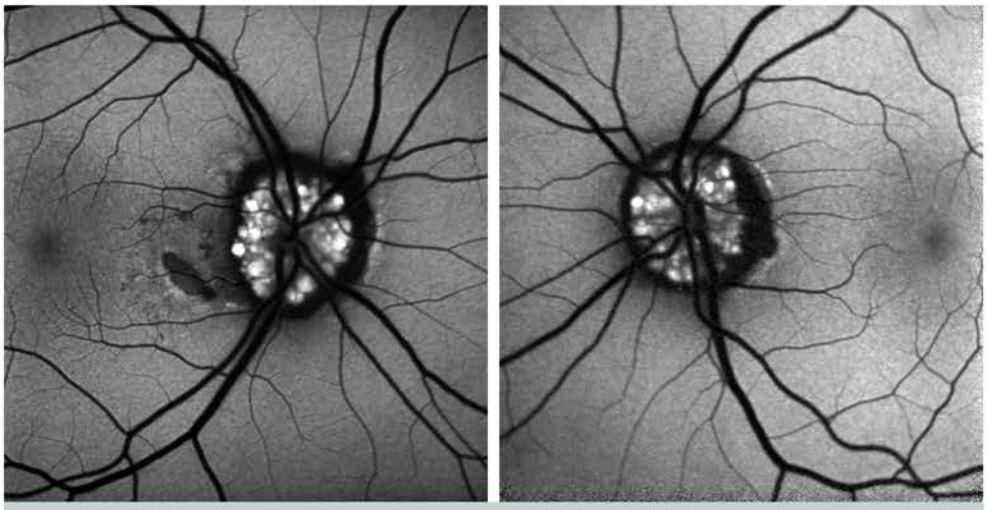
Introduction:

An asymptomatic 35-year-old white male was referred to our clinic for retinal and optic nerve evaluation. Specifically, the patient denied any headaches, pulsatile tinnitus, or transient visual obscurations.

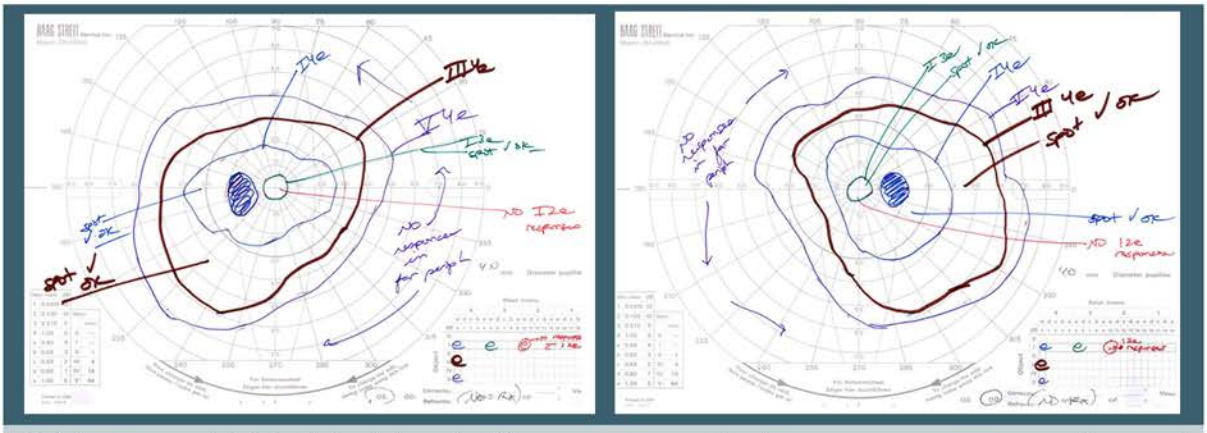


Fundus autofluorescence photos of the right and left eyes, respectively, demonstrating a "lumpy-bumpy" appearance to the optic discs with focal spots of hyperfluorescence. The right eye also shows peripapillary hypofluorescence most likely corresponding to an old choroidal neovascular membrane.

Exam:

His vision was 20/20 in both eyes with normal intraocular pressures and anterior chamber exams. His fundus exam was remarkable for optic disc head elevation of both eyes without vessel obscuration or hemorrhage. The right eye additionally had chorioretinal scarring abutting the temporal optic nerve but not encroaching the fovea. Optical coherence tomography (OCT) of his maculae showed mild maculoschisis of both eyes temporally without fluid overlying the peripapillary scar/pigment epithelial detachment of the right eye, and a nerve fiber layer analysis of his optic nerves demonstrated inferotemporal thinning of the right eye and mild-to-moderate diffuse thinning of the left eye.

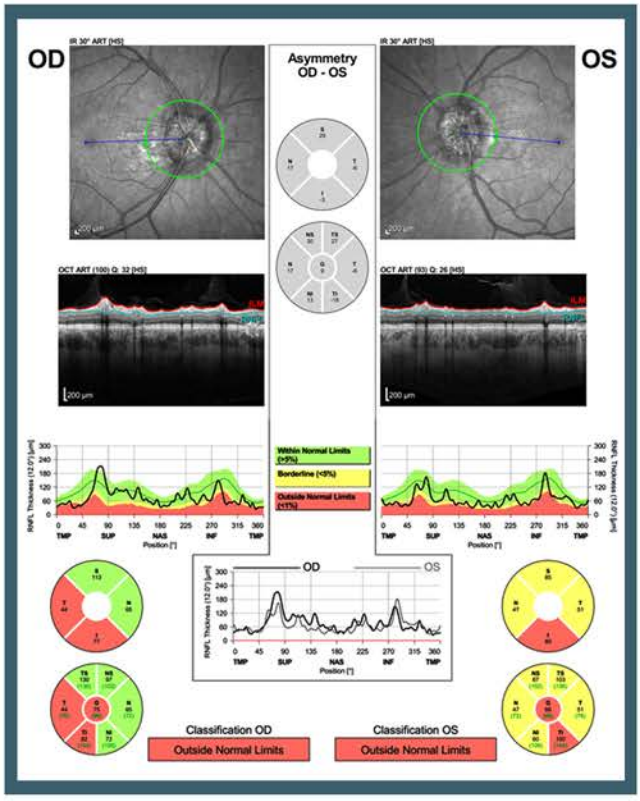
Fundus autofluorescence photos showed numerous hyperfluorescent foci within each optic nerve head along with an area of hypofluorescence surrounded by a ring of hyperfluorescence in the right eye aligning with the aforementioned chorioretinal scar. Ultrasound exam revealed hyperechoic optic nerve heads and were otherwise unremarkable. Finally, Goldmann visual fields demonstrated mirrored inferonasal field loss with perhaps an enlarged blind spot of the left eye but without obvious



Goldmann visual fields of the left and right eyes, respectively, demonstrating symmetric inferonasal constriction and perhaps an enlarged blind spot in the left eye.

arcuate scotomata or hemifield cut.

In sum, the above findings are most consistent with maculoschisis of both eyes, optic disc drusen (ODD) of both eyes, and an inactive peripapillary choroidal neovascular membrane (CNVM) of the right eye. The remainder of this case presentation will focus on the latter two diagnoses.

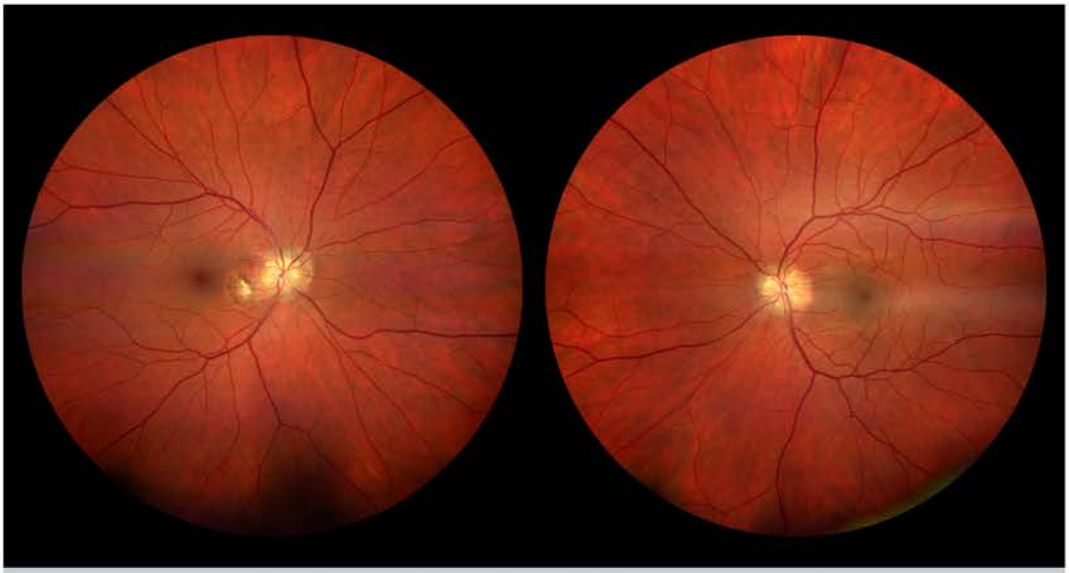


Retinal nerve fiber layer analysis of the right and left eyes, respectively, showing thinning left worse than right.

Discussion:

ODD is a common finding, especially in the Caucasian population, occurring in at 0.4 to 3.7% of individuals, usually bilaterally. There is no proven genetic basis for ODDs though they seem to follow a dominant inheritance pattern with incomplete penetrance. They can occur either within the head of the optic nerve or are buried, with the latter likely leading to an underreporting of cases. They are composed of acellular material and are most striking when calcified, though not all ODD is. The pathogenesis is thought to stem from abnormal axonal metabolism of the optic nerve, leading to extrusion (and sometimes calcification) of intracellular material. This extrusion can then lead to the characteristic "lumpy-bumpy" appearance of nerve head drusen that is sometimes difficult to differentiate from papilledema. Many patients with ODD are asymptomatic, but this does not mean that the disease process is benign. The drusen can cause symptomatic visual field defects, CNVM formation, hemorrhage, and ischemic optic neuropathy. It is unclear in our case if the maculoschisis is an incidental finding or related to the disc drusen. It is possible the cystic spaces found actually relate to transsynaptic degeneration as a result of compressive injury from the disc drusen, though the literature fails to reveal any definite correlation between the two diagnoses.

ODD is easiest to recognize when calcified, visible on ultrasound scan or autofluorescence imaging Cross-sectional OCT of the nerve head can also clearly demonstrate the drusen. Computed tomography may catch calcified ODD, though their use as a screening tool is discouraged due to poor spatial resolution and exposure to radiation. The drusen can grow with age and lead to progressive vision loss. It is important to differentiate the nerve head elevation from ODD (pseudopapilledema) from true papilledema, a swelling of the optic nerves caused by increased intracranial pressure. This can sometimes be challenging, as both can present with vision and vascular changes (including peripapillary hemorrhage) and patients can have both diagnoses simultaneously. Thus, in symptomatic patients (especially those with headache or other neurologic symptoms), it is reasonable to obtain brain imaging and/or lumbar puncture



Color photos of the right and left eyes, respectively.

even in the presence of demonstrable ODD.

There is no clear-cut consensus on treatment strategy for isolated ODD at this time. Most clinicians follow these patients with serial visual field and nerve fiber layer thickness measurements, and some will start therapy to lower intraocular pressure if suspicious of progressive neurodegeneration as glaucoma can occur simultaneously with ODD. Treatment strategies to improve ocular perfusion, either medically or with nerve decompression, are unproven. Finally, active peripapillary CNVMs can either be observed or treated with injection of anti-VEGF agents. We tend to be more aggressive with treatment of temporal CNVMs as these are more likely to be fovea threatening.

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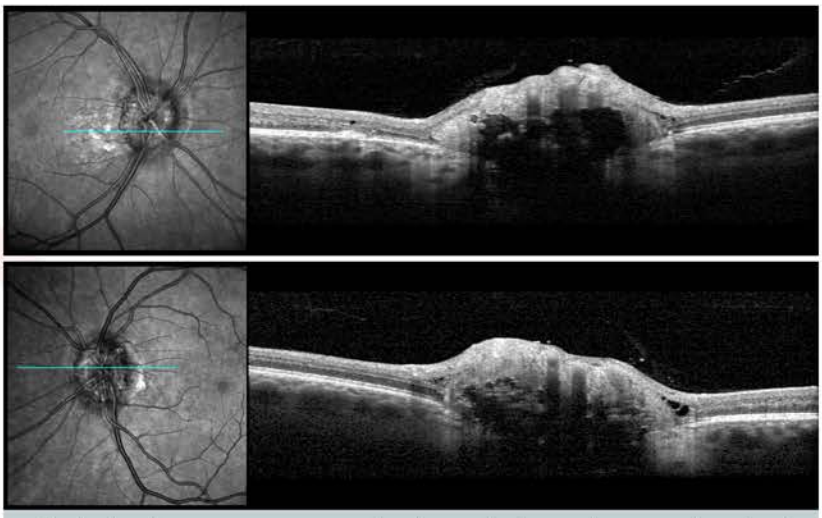
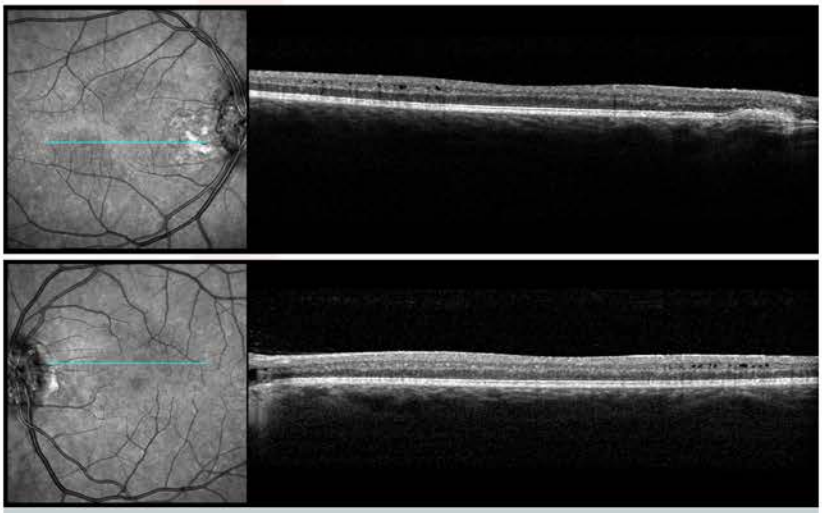
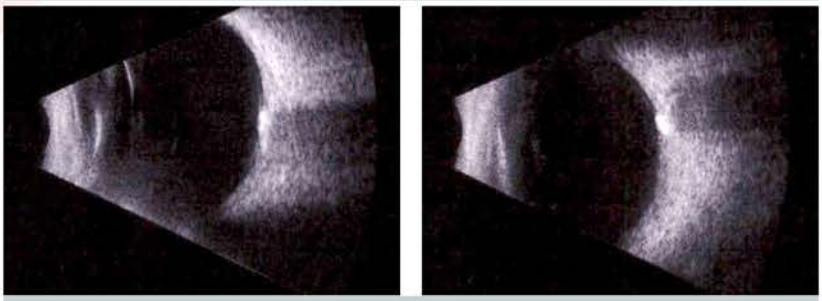


Optical coherence tomography through the optic nerve heads of the right (top) and left (bottom) eyes demonstrates cross-sectional irregularity of the nerves with a focal area of outer retinal atrophy in the right eye.



Optical coherence tomography of the right (top) and left (bottom) eyes showing mild temporal bilateral maculoschisis and a fibrovascular peripapillary disruption of the right eye without fluid.



Ultrasound photos of the right and left eyes, respectively, demonstrating hyperfluorescence of the optic nerve heads.

Case of the Month Supported by:

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