Colobomas of the Optic Nerve

52-year-old woman presented for a routine eye exam with no visual complaints. Her best-corrected visual acuity was 20/20 in both eyes. Funduscopy of the right eye (Fig. 1) revealed a massive optic nerve coloboma with no other associated abnormalities on exam. Funduscopy of the left eye (Fig. 2) revealed a much smaller optic nerve coloboma with no other associated abnormalities.

Colobomas of the optic nerve usually occur bilaterally. They are frequently associated with defects in the *PAX6* gene. Serous detachments of the macula are also frequently observed in patients with this condition. Retinal vessels can be anomalous as well, in that they are often increased in number and have a generally straight course in the peripapillary region. Visual prognosis can be poor when other ocular developmental abnormalities such as microphthalmos are present, but people with isolated optic nerve colobomas without other ocular malformations can have excellent visual status, as this case demonstrates.

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