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An Atypical Optic Nerve Head Mass

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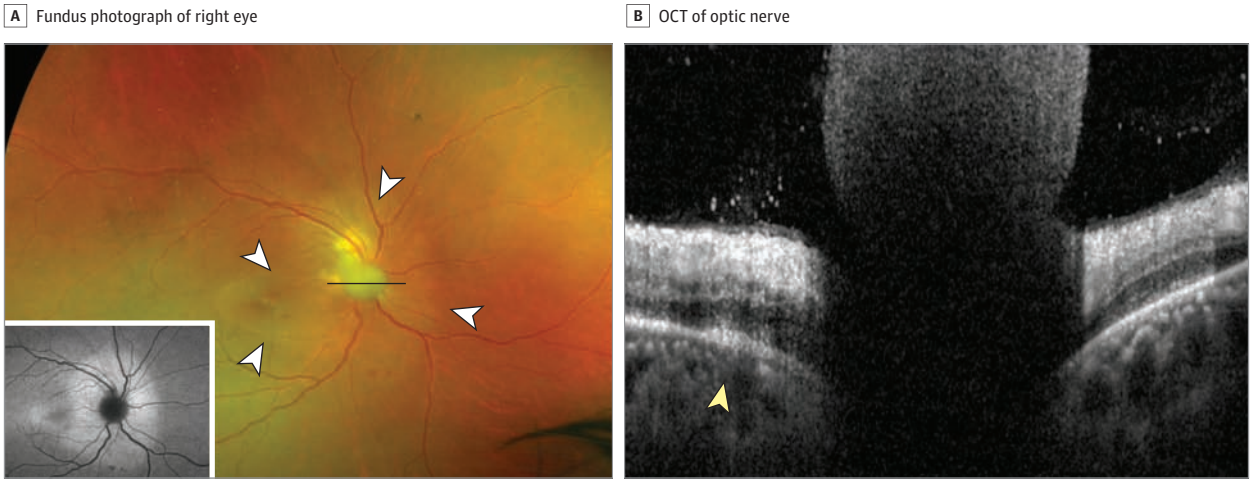

Figure 1. Fundus photograph (A) of the right eye showing vitreous haze, an optic nerve head mass, and a subtle posterior pole yellow/white placoid lesion (white arrowheads) with peripheral dot blot hemorrhages; autofluorescence imaging (left lower corner insert) delineates the posterior placoid lesion. Optical coherence tomography (OCT) (B) of the optic nerve (black line corresponding to cross-sectional area) shows a large hyporeflective optic nerve head mass and ellipsoid zone dysfunction (yellow arrowhead). The anterior portion of the optic nerve head gumma is outside the single optical coherence tomography B-scan window.

A 47-year-old man presented to our clinic with a sudden decrease in vision in the right eye. One month prior, he had presented to the emergency department with binocular diplopia, bilateral cranial nerve 6 palsies, and an undifferentiated pontine mass on brain magnetic resonance imaging. Broad serologic workup results were negative, except for a positive syphilis total antibody with a nonreactive rapid plasma reagin test. Lumbar puncture with flow cytometry and cerebrospinal fluid venereal disease research laboratory test (CSF VDRL) result was also negative. The patient reported a history of syphilis treated with intramuscular penicillin 9 years prior. He was treated with systemic corticosteroids and discharged home on a steroid taper.

On our examination, his visual acuity was count fingers in the right eye and 20/20 in the left. He had a right afferent pupillary defect and intraocular pressures were 12 in the right eye and 11 in the left. Anterior segment examination was normal. Posterior examination of the right eye revealed 3+ vitreous cell, 2+ vitreous haze, and a large optic nerve head mass with optic disc hyperemia, scattered dot hemorrhages, and a subtle placoid lesion in the posterior pole (**Figure 1**A). Optical coherence tomography of the optic nerve demonstrated a large spherical mass protruding out of the optic nerve head (Figure 1B) and granular disruption of the ellipsoid zone in adjacent peripapillary retina.

WHAT WOULD YOU DO NEXT?

A. Perform diagnostic anterior chamber and/or vitreous tap and inject vancomycin and ceftazidime

B. Perform pars plana vitrectomy for diagnostic vitreous biopsy

C. Reinitiate systemic prednisone therapy

D. Obtain additional treponemal test and, if positive, begin intravenous penicillin

CME Quiz at jamacmelookup.com

Diagnosis

Syphilitic gumma of optic nerve head and presumed neurosyphilis

What to Do Next

D. Obtain additional treponemal test and, if positive, begin intravenous penicillin

Discussion

A focused differential diagnosis for unilateral unpigmented optic nerve head mass includes tumors, such as lymphomas and astrocytomas; infections, such as syphilis and tuberculosis; and inflammatory etiologies, such as sarcoidosis. This patient presented with clinical findings highly suggesting of syphilis (placoid lesion and optic nerve head mass). However, preliminary serologic evaluation for

syphilis demonstrated discordant results with a positive treponemal syphilis total antibody test result and a nonreactive rapid plasma regain test result. A second confirmatory treponemal test result (treponema pallidum particle agglutination) was positive, suggesting either previously treated syphilis or tertiary syphilis. In the context of the ocular and neurologic findings, intravenous penicillin was initiated (choice D).

Obtaining a fluid sample is a possibility; however, the optic nerve head mass would be atypical for bacterial or fungal endophthalmitis (choice A). A pars plana vitrectomy would be too invasive at this stage (choice B). Initiating oral prednisone in the setting of active infection would exacerbate the underlying syphilitic process (choice C).

Discordant syphilis test results may require a third test (commonly treponema pallidum particle agglutination) to act as a tiebreaker in certain situations. In this case, the clinical presentation and serologic results likely represent late (tertiary) syphilis as opposed to reinfection, given the negative rapid plasma reagin test result and observed gumma (granuloma surrounding *Treponema pallidum* spirochetes). Overall, the robust vitritis, posterior pole placoid lesion, and multiple gummatous lesions (optic nerve head and pons) in this patient are consistent with active ocular and neurosyphilis.[1]

Despite nonreactive nontreponemal serologic and CSF VDRL testing, this patient was found to have neurosyphilis. The accuracy of nontreponemal testing in neurosyphilis is lower when compared to primary or secondary syphilis, with sensitivity ranging from 49% to 87.5% and specificity from 74% to 100%.[2] A large series[3] examined the reactivity of CSF VDRL, fluorescent treponemal antibody, and fluorescent treponemal antibody absorption tests in 150 patients with syphilis and found that 22.2% of neurosyphilis cases had a reactive CSF VDRL, suggesting a limitation in relying solely on CSF VDRL for diagnosing neurosyphilis.

Ocular syphilis can be difficult to diagnose, given its ability to masquerade as different clinical entities, such as dacryoadenitis, interstitial keratitis, or even retinal vasculitis.[4] It can affect the optic nerve as perineuritis, optic neuritis, or papillitis, characterized by optic nerve swelling with possible peripapillary hemorrhages and, rarely, venous engorgement.[5] Neurosyphilis often intersects with ocular syphilis,[6] and syphilitic gummas can arise throughout the central nervous system in focal areas of syphilitic meningitis, more often in early neurosyphilis.[6]

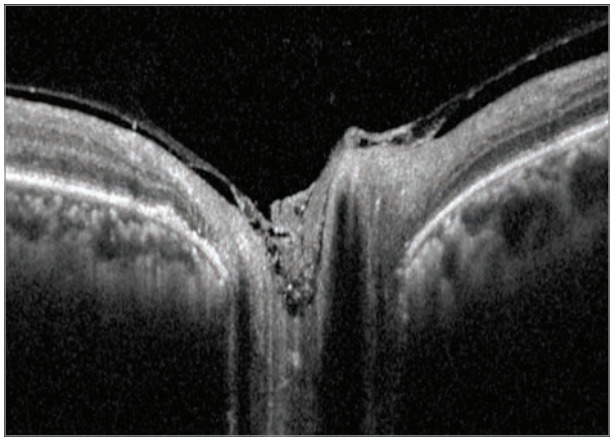

Figure 2. Optical coherence tomography of the optic nerve head showing resolution of optic nerve head gumma and improvement of the peripapillary ellipsoid zone.

Gummas of the optic nerve are rare.[5,7] A recent report demonstrated a case of a presumed optic nerve head gumma, given its fluorescein pattern and prominent telangiectatic vessels with elevation of the lesion on optical coherence tomography.[5] The patient had complete resolution of the optic neuropathy after monotherapy intravenous penicillin, supporting the diagnosis.

Patient Outcome

After 14 days of intravenous penicillin, the patient's vision improved to 20/40. The disc edema and optic nerve head gumma resolved, and there was reconstitution of the ellipsoid zone (**Figure 2**).

ARTICLE INFORMATION

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