Thiamine Deficiency Optic Neuropathy

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Introduction

A rare cause of vision loss in developed countries is nutritional optic neuropathy. The typical presentation includes painless, bilateral, symmetric vision loss, central or cecocentral scotoma, and reduced color vision. Fundoscopy is often normal early in the disease course and may eventually demonstrate optic nerve pallor. The most common etiologies including vitamins B1, B2, B3, B6, B9, B12, and Copper (1). A nutritional optic neuropathy should be suspected in patients with limitations in their diet, such as in chronic, frequent alcohol use, or in those who many not absorb nutrients effectively, such as those with bariatric surgery, chronic diarrhea, pernicious anemia, etc (2).

Case Presentation

- 39 year old female with no past ocular disease
- Presents with central visual loss, difficulty seeing colors, and tingling of her lower extremities which progressed slowly over multiple months.
- Increased alcohol consumption over last four years; currently drinking five glasses of wine per day.
- Ophthalmic exam reveals visual acuity 20/400 s.c. bilaterally and temporal optic nerve pallor (figure 1); other wise unremarkable.
- MRI brain and orbits with mild atrophy of the cortex more than expected for age, otherwise unremarkable.
- Ocular coherence tomography (OCT) - macula with generalized thinning bilaterally.
- OCT - Retinal Nerve Fiber Layer (RNFL) with superior and inferior thinning of the right optic nerve and temporal and inferior thinning of the left optic nerve (figure 2).
- Humphrey Visual Field 24-2 with central scotoma bilaterally (figure 3).
- Vitamin panel sent – thiamine (Vitamin B1) deficient 54.2nmol/L (normal 66.5-200nmol/L).
- Patient started on thiamine replacement intramuscularly and then 100mg daily.
- Three months following initiation of thiamine replacement, vision improved to 20/200 OD s.c., 20/150 OS s.c. and there was a drastic improvement of her visual fields (figure 3).

Testing

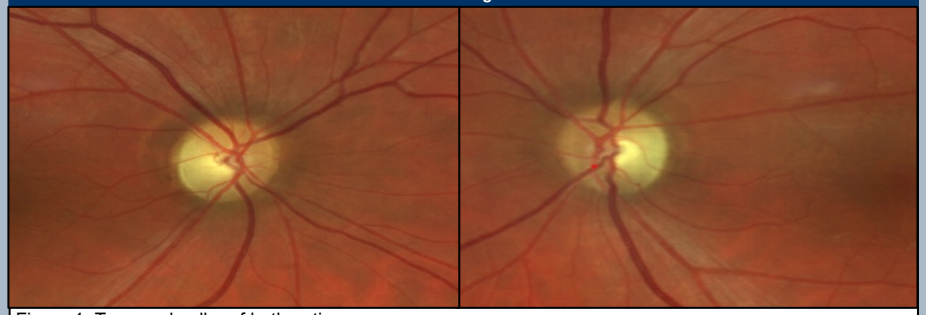

Figure 1. Temporal pallor of both optic nerves

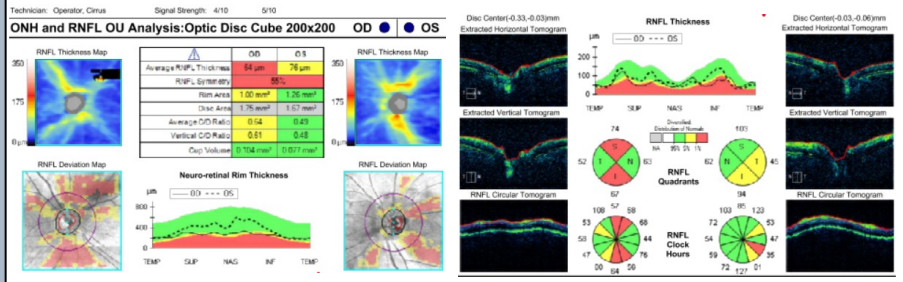

Figure 2. OCT-RNFL demonstrating bilateral optic nerve thinning

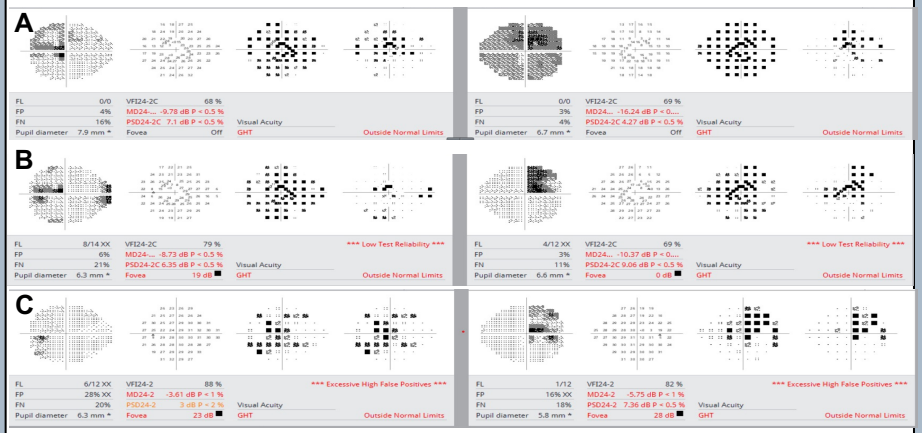

Figure 3. A. Humphrey Visual Field (HVF) 24-2 at original presentation. B. HVF one month after initiating thiamine replacement. C. HVF three months following initiation of thiamine replacement.

Discussion

Nutritional optic neuropathies remain a challenging diagnosis as their discovery relies largely on history and not on ophthalmic exam. Visible sequelae from this disease often develop late in its course. It often coincides with other causes of optic neuropathy such as toxic or hereditary causes and thus the differential should remain broad on initial assessment and include a full neurologic evaluation, brain and orbit imaging, lab work for toxic sources and nutritional deficits, and possible genetic testing (1). The mechanisms of nutritional optic neuropathies have not been fully elucidated, however the common pathway seems to be linked to oxidative stress (3). Prognosis is variable and in general is more favorable when treatment is initiated as early as possible (4).

Conclusion

- Nutritional optic neuropathy remains a challenging diagnosis due to its rarity in first world countries and its reliance on adequate history taking given a typical unremarkable ophthalmic exam early in its course.
- Common nutritional deficits that should be tested include vitamins B1, B2, B3, B6, B9, B12, and copper.
- Patients can have improvements in their visual function after initiating treatment.

References

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