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Vision Loss in a Teenage Girl With Postconcussion Syndrome

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A girl in her teens with a history of postconcussion syndrome was referred for retinal evaluation after failing her vision examination for her learner's permit for driving. She complained of progressively declining, blurred vision for 2 years. She denied floaters, flashes of light, headaches, and eye pain. The patient had a concussion 1 year prior after becoming dehydrated during marching band practice, fainting, and hitting her head on pavement. Her postconcussion symptoms included headaches, dizziness, and dysarthria. The headaches and dizziness resolved but the dysarthria remained. Brain magnetic resonance imaging (MRI) within the last year was normal. She denied acute vision changes after the concussion. Family history was unremarkable for vision loss, glaucoma, inherited retinal degeneration, and autoimmune disease.

Best-corrected visual acuity was 20/80 OU. Ishihara testing revealed red-green color deficiency bilaterally. Intraocular pressures, extraocular motility, confrontational visual fields, and pupils were normal. Anterior segment examination was unremarkable. Ophthalmoscopic examination demonstrated mild mottled retinal pigment epithelial abnormalities bilaterally (Figure, A).

Fundus autofluorescence showed bilateral mild macular hyperautofluorescence likely within normal range without evidence of flecks. Retinal spectral-domain optical coherence tomography demonstrated bilateral disruption of the interdigitation and ellipsoid zones subfoveally (Figure, B). Electroretinogram (ERG) revealed delayed and decreased amplitude responses to 30-Hz flicker with normal dark-adapted responses. Goldmann visual field testing showed mild bilateral nasal constriction.

Two months after presentation, the patient returned with new balance difficulties and worsening dysarthria. Horizontal and vertical saccades were markedly slowed bilaterally.

Diagnosis

Spinocerebellar ataxia 7

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What to Do Next

B. Genetic testing

Discussion

When the patient returned with worsening dysarthria, saccadics slowing, and balance difficulties, the differential diagnosis narrowed to conditions producing both slowly progressive vision loss and neurological findings. A unifying diagnosis was sought.

A second brain/orbital MRI with gadolinium and lumbar puncture to provide cerebrospinal fluid for an oligoclonal band screen would aid in diagnosing multiple sclerosis (MS). Multiple sclerosis is a common cause of combined vision loss and focal neurological symptoms in young women. However, several factors make MS unlikely. The patient had a normal brain MRI within a year of her visit. Additionally, vision loss in MS is typically acute and occurs over days or weeks rather than progressively over years.¹ While saccades can be slow in MS, such as from internuclear ophthalmoplegia, vertical saccades are rarely affected. An audiogram is useful in diagnosing Usher syndrome. The type III subtype can produce progressive vision and hearing loss with balance problems presenting in adolescence.^{2,3} The vision loss in Usher syndrome, though, initially affects rod photoreceptor function. The ERG showed normal scotopic function.³ Usher syndrome would also not produce the foveal changes found on spectral-domain optical coherence tomography.³

Instead, the patient's dysarthria, balance problems, saccadic slowing, and slowly progressive vision loss associated with cone dystrophy prompted genetic testing for spinocerebellar ataxia 7 (SCA7). Genetic testing demonstrated 65 CAG repeat expansions in *ATXN7*, consistent with SCA7. Spinocerebellar ataxia 7 is an autosomal dominant subtype of the SCAs.⁴⁻⁷ The expansions cause neuronal dysfunction, leading to cell loss in the retina, cerebellum, and brainstem. An inverse correlation exists between the number of CAG repeats and age at onset.⁴⁻⁶ The symptoms in turn depend on the age at presentation, and earlier onset portends faster disease progression. In infancy and childhood (first decade of life), symptoms include muscle wasting, failure to thrive, and loss of motor milestones.⁸ In adolescent- and adult-onset cases such as this one, presentation includes ataxia, dysarthria, and dysmetria.⁴ Importantly, though the disease is inherited in an autosomal dominant pattern, a negative family history does not preclude the diagnosis. A parent may remain asymptomatic or present after an affected child due to genetic anticipation (especially in paternal transmission).^{5,6}

In both early- and late-onset cases, visual symptoms include decreases in central visual acuity, defects in color vision, slow saccades, and photophobia.^{5,6} Progression to blindness occurs earlier with childhood onset. Visual symptoms may occur before, during, or after the presentation of neurological findings.⁶ Therefore, initial presentation may be limited to ophthalmologic findings with a normal brain MRI. Saccadic slowing may be subtle and requires thorough motility testing as global saccadic slowing points to SCA7.

Retinal degeneration is present in almost all patients with SCA7 and differentiates this subtype from the other SCAs. Cone cell degeneration occurs before involvement of rod cells, and ERG reveals cone cell dysfunction.⁷ Symptoms reflecting cone cell degeneration such as deficits in color vision and abnormal ERG readings may precede retinal degeneration on examination as this case demonstrates.⁷ On ophthalmoscopy, early changes include subtle arteriolar attenuation and loss of the foveal light reflex.⁹ With disease progression, atrophic macular changes develop along with pigmentary changes.⁹ As retinal degeneration advances, rod photoreceptor cells also deteriorate leading to complete blindness.^{7,9} However, as in this patient, early ophthalmoscopy may be normal even with vision abnormalities.

Patient Outcome

There is no curative treatment for SCA7. Vision loss is progressive. Neurological dysfunction often leaves patients wheelchair-bound. Dysphagia presents a risk of aspiration. In association with occupational, physical, and speech therapy, the patient has continued to attend high school. The patient and her family were referred for additional genetic counseling.

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WHAT WOULD YOU DO NEXT?

- A.** MRI of the brain/orbits with contrast
- B.** Genetic testing
- C.** Lumbar puncture
- D.** Audiogram

Figure.

Right eye multicolor fundus image and retinal spectral-domain optical coherence tomography (SD-OCT). A, Multicolor fundus image demonstrates mild macular mottling. The optic disc and retinal vessels are unremarkable. B, Retinal SD-OCT shows normal foveal contour with disruption and loss of the interdigitation and ellipsoid zones subfoveally.