Birdshot retinochoroidopathy in the absence of typical chorioretinal lesions

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Abstract
A patient with Birdshot retinochoroidopathy that presented with isolated optic nerve swelling is reported. Neuroimaging and serum testing for infectious or inflammatory etiologies were unremarkable. Indocyanine angiography revealed multiple round dark fundus lesions in both eyes. This raised suspicion for Birdshot retinochoroidopathy, which was confirmed with genetic testing. Ocular inflammation resolved following treatment with mycophenolate mofetil and a dexamethasone intravitreal implant.

Key words: retinochoroidopathy; diagnosis; management.

Introduction
Birdshot retinochoroidopathy (BSRC) is an ocular inflammatory disease characterized by depigmented lesions radiating from the optic disc. It may, however, present with isolated optic disc edema, vitritis, or vasculitis. ICG can play a critical role in diagnosing BSRC in cases where characteristic fundus lesions are subtle or absent.

We describe a case of Birdshot retinochoroidopathy that presented with isolated optic nerve swelling. Neuroimaging and serum testing for infectious or inflammatory etiologies were unremarkable.

Figure 1: Left optic nerve (top left) at initial presentation showing disc edema, hemorrhages and cotton wool spots. HVF (top right) was normal OD and showed nonspecific shallow defects OS. Bilateral fundus photos (bottom) after short trial of systemic corticosteroids.
Indocyanine angiography (ICG) revealed multiple round fundus lesions in both eyes. This raised suspicion for BSRC, which was confirmed with genetic testing. Ocular inflammation resolved following treatment with mycophenolate mofetil (MMF) and a dexamethasone intravitreal implant (Ozurdex®; Allergan Inc., Irvine, CA).

Case Report
A 63-year-old woman with a history of Hashimoto’s thyroiditis presented to neuro-ophthalmology for evaluation of left optic nerve swelling and left sided proptosis. She described floaters in both eyes for several weeks. On examination, her visual acuity (VA) was 20/20 OU. Her pupils and color vision were normal. She had 2 mm of proptosis and optic disc edema with associated hemorrhage and cotton wool spots in the left eye (OS) (Figure 1). Her Humphrey visual field was normal in the right eye (OD) and showed a few shallow nonspecific defects in the OS (Figure 1). Her orbital MRI was normal. One month later, her examination was stable, except for a new vitritis in the OS. Laboratory work up included serum testing for ANCA, ACE, ANA, RPR, QuantiFERON-TB Gold In-Tube assay (QFT-GIT), and Lyme all negative.

She was prescribed a two-week burst of oral prednisone. Two months later she was seen in the retina clinic. Her visual acuity was 20/25 OU. She had improvement of the optic disc edema, but she had developed vitritis in both eyes. The choroidal vessels appeared prominent (Figure 1).

Fluorescein angiography (FA) showed leakage of the optic nerves and vessels OU and macula OS. The ICG revealed multiple dark small round hypofluorescent lesions in the posterior pole and mid periphery (Figure 2). Macular OCT was normal OU. Extramacular enhanced depth optical coherence tomography (EDI) showed hyporeflective areas of choroidal depigmentation (arrows) surrounded by choroidal vessels (Figure 3). These findings raised suspicion for BSRC.

She was HLA A29 positive, consistent with the diagnosis of BSRC.

She was started on cyclosporine 125 mg/day and prednisone 40 mg/day. Follow up over a period of four months showed marked improvement in symptoms, with resolution of vitritis and optic disc edema OU. The FA was normal. Because of significant side effects from cyclosporine and prednisone, the medication was switched to MMF. Recurrence of
Inflammation developed while on MMF and she underwent dexamethasone intravitreal implants OU, which successfully controlled her inflammation for several months.

**Comments**

Birdshot retinopathy, a bilateral ocular inflammatory disease, most frequently affects middle-aged Caucasians. Typical findings include cream-colored round or oval shaped lesions at the level of the retinal pigment epithelium and choriocapillaris. Of all uveitides, BSRC has the strongest class 1 major histocompatibility antigen association. Up to 98% of patients with BSRC are positive for HLA A29, compared to only 7% in the general population.

Gupta et al. reported a single patient with BSRC in which the diagnosis was made without clinical lesions. In four other cases, patients experienced significant delay in diagnosis because retinal lesions did not develop until 2-8 years after symptom onset.

The case herein initially presented as unilateral optic disc edema. We were able to diagnose BSRC even though the characteristic fundus lesions were not observed. The classic findings were detected with ICG, which prompted HLA typing.

Our case illustrates several important features: 1) BSRC can present as unilateral disc edema in absence of other symptoms; 2) FA can identify occult vasculitis among patients with unexplained vitritis; 3) ICG can reveal BSRC lesions not otherwise apparent on clinical exam or other imaging modalities; 4) EDI may demonstrate hyporeflective spots surrounded by choroidal vessels, as noted in previous studies of BSRC. It would have been interesting to see how an ICG, OCT EDI and the FA would looked at initial presentation, and if these tests could have facilitated an earlier diagnosis of BSRC before the vitritis occurred.

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**REFERENCES**


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Figure 3: The extra macular EDI-OCT showed hypo reflective spots possibly due to choroidal pigmenary changes or inflammatory cellular infiltrates (arrows) surrounding some prominent choroidal vessels.