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Case report

HLA-A29 negative Birdshot-like chorioretinopathy associated with common variable immunodeficiency

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ABSTRACT

Purpose: To report the longest ophthalmic follow-up and the associated ocular complications of HLA-A29 negative Birdshot-like chorioretinopathy (BLCR) associated with common variable immunodeficiency (CVID).

Observations: A 22-year-old man known for CVID presented with a 3-month history of decreased visual acuity OS. Funduscopy revealed significant cystoid macular edema OS, as well as optic disk edema and chorioretinal infiltrates without signs of vitritis OU. No infectious, inflammatory or neoplastic etiologies were identified. He subsequently received one dose of intravitreal triamcinolone OS which completely resolved the macular edema. The optic nerve edema persisted despite the addition of intravenous immunoglobulin. His visual acuity was 20/20 OU at the 24th follow-up month.

Conclusion: and importance: To our knowledge, this is the third case of HLA-A29 negative BLCR associated with CVID. It is the first case with long-term follow-up providing, in consequence, the best understanding of the natural history and possible complications of this rare disease. Aggressive systemic treatment, in collaboration with an immunologist, is generally needed to control the ophthalmic complications.

1. Introduction

Common Variable Immunodeficiency (CVID) is a primary immunodeficiency leading to defects in B-cell differentiation and a subsequent decrease in antibody-producing plasma cells along with hypogammaglobulinemia. Patients with this disorder typically present with recurrent sino-pulmonary bacterial infections as well as increased risk for autoimmune disease.[1–3] In North America, CVID is the most common primary immunodeficiency and the most severe form of antibody deficiency affecting both children and adults.[4] The etiology of CVID remains unclear.

Birdshot chorioretinopathy (BCR) is a rare autoimmune chorioretinal disease manifesting as bilateral posterior uveitis with typical white-creamy hypopigmented choroidal lesions. Its pathogenesis remains poorly understood despite its strong association with the HLA-A29 haplotype, suggesting a possible pathophysiological role for T-cells. Clinically, most patients with BCR present with blurred vision, floaters, nyctalopia and a typical fundus appearance.[5,6]

A literature review detected nine cases of CVID that manifested with unique ocular findings during the course of disease. Among these, HLA-A29 negative Birdshot-like chorioretinopathy (BLCR) was reported in only two cases.[7–12] In this work, we report a unique case of chorioretinitis presenting with creamy chorioretinal infiltrates with a scattering mimicking that of BCR (hence, the “birdshot-like” naming) in a patient with CVID. To our knowledge, we report the third case of HLA-A29 negative BLCR associated with CVID, and we document, for the first time, its natural history over a 2-year follow-up period.

2. Case report

A 22-year-old man was referred to our clinic after reporting a 3-month history of decreased vision in his left eye. His past medical history was relevant for CVID and psoriasis. The CVID was diagnosed by the allergy and immunology services after presenting with recurrent sino-pulmonary infections and frequent otitis media in his childhood. At that time, a diagnosis of CVID was made based on marked hypogammaglobulinemia (IgG and IgA). The patient was subsequently treated with subcutaneous immunoglobulins.

The patient, who was known for −7.5 D of myopia OU, did not report any significant past ocular history. His visual acuity (VA) at presentation was 20/30 OD and 20/60 OS. Pupil reaction to light was normal. His intraocular pressures were 12 mmHg OU. The anterior segment examination was also within normal limits and demonstrated no cellular reaction. Fundoscopy revealed bilateral nasal chorioretinal creamy infiltrates as well as bilateral optic nerve edema more pronounced in the left eye (Fig. 1A and B). Fluorescein angiography (FA)

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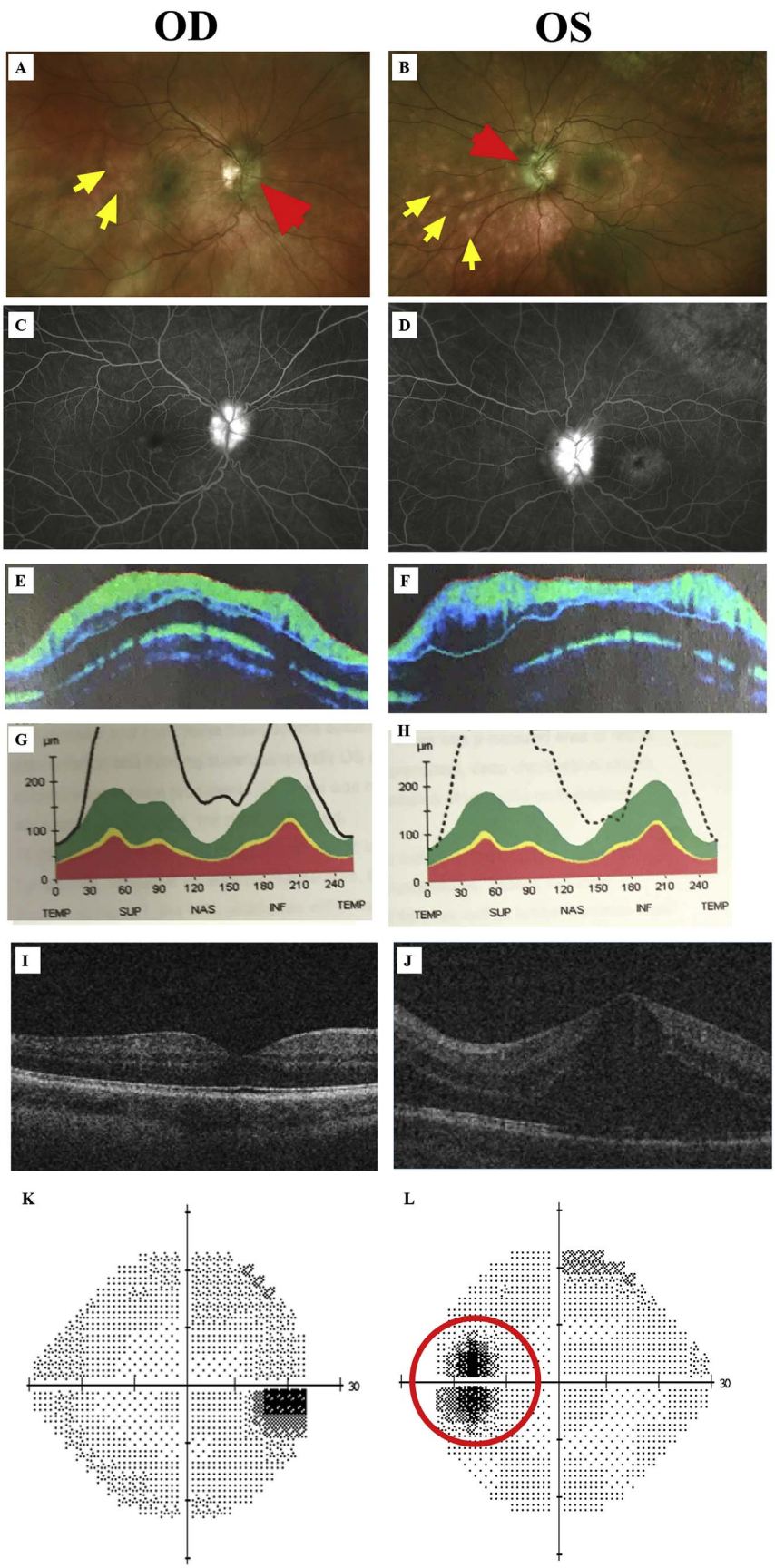

Fig. 1. Initial presentation: 22-year-old man with HLA-A29 negative Birdshot-like chorioretinopathy associated with common variable immunodeficiency. A & B. Fundus photo of OD and OS, respectively, showing optic nerve edema (red arrow) and yellow creamy chorioretinal infiltrates (yellow arrows). **C & D.** Mid phase fluorescein angiography (FA) showing diffuse optic nerve hyperfluorescence OD and diffuse optic nerve hyperfluorescence as well as central foveal leakage in a petalloid pattern OS, respectively. **E & F.** Optic nerve optical coherence tomography (OCT) showing significant optic nerve elevation due to edema OD and OS, respectively. **G & H.** Retinal nerve fiber layer (RNFL) thickness graph showing increased thickness due to edema in OD and OS, respectively. **I & J.** Macular OCT showing normal retinal architecture OD and significant cystoid macular edema (CME) OS, respectively. **K & L.** Humphrey visual field (30-2 SITA) showing no abnormalities OD and enlarged blind spot (red circle) OS, respectively. (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)