

Diffuse Placoid Choroidopathy in a Patient with Common Variable Immunodeficiency

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Established Facts

- Common variable immunodeficiency syndrome can be complicated by lymphadenopathy and granulomatous disease.

Novel Insights

- The eye manifestations resemble granulomatous or infiltrative disease such as lymphoma, but may actually be a manifestation of benign lymphoproliferative disease.
- These findings may easily be misdiagnosed, and awareness of ocular manifestations of common variable immunodeficiency is beneficial, and may prevent unnecessary biopsy of these ocular lesions.

Key Words

Choroidal granuloma • Common variable immunodeficiency • Hypogammaglobulinemia • Placoid choroidopathy • Sarcoidosis

Abstract

Unilateral diffuse placoid choroidopathy can be seen in the setting of common variable immunodeficiency (CVID). A 55-year-old woman with a 20-year history of CVID presented with multiple hypopigmented placoid lesions in the right eye. Diffuse lymphadenopathy led to lymph node biopsies

(mediastinal and axillary) and angiotensin-converting enzyme and serum calcium level determination, which were all normal. CVID, the most common type of significant hypogammaglobulinemia, may be associated with a placoid choroidopathy. The cause of this CVID choroidopathy is unclear at this time, but may be secondary to lymphoid hyperplasia or granulomatous disease. Copyright © 2008 S. Karger AG, Basel

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Introduction

Common variable immunodeficiency (CVID) is the most common significant type of hypogammaglobulinemia. It is a rare disorder characterized by defects in antibody synthesis. CVID typically presents with recurrent bacterial infections, generalized lymphadenopathy, and splenomegaly. Ocular involvement is rarely reported. This report describes a patient who developed unilateral placoid choroidopathy after a 20-year history of CVID.

Case Report

A 55-year-old Caucasian woman presented for a second opinion regarding a previous diagnosis of birdshot chorioretinopathy. At the time of diagnosis, she was treated with prednisone for 2 weeks. Her symptoms diminished, but residual intermittent blurred vision remained. HLA-A29 testing was negative.

The patient's medical history was remarkable for a 20-year history of CVID with typical complications of the disease including splenomegaly and subsequent splenectomy, chronic generalized lymphadenopathy, idiopathic thrombocytopenia purpura, and bronchiectasis. The patient also reported multiple episodes of shingles affecting the periocular skin of the right eye, the last occurring approximately 2.5 years prior to presentation.

On examination, visual acuity was 20/25 in the right eye and 20/20 in the left eye. Intraocular pressures and anterior segment examination were unremarkable. Fundus examination of the right eye revealed poorly demarcated hypopigmented lesions surrounding the posterior pole and extending to the mid-periphery. In the central macula, an irregular pattern of pigment clumping was noted (fig. 1). There were no vitreous cells and no evidence of previous herpetic retinal infection. The left eye was completely unremarkable.

Computed tomography showed diffuse lymphadenopathy. Positron emission tomography indicated hypermetabolic adenopathy suspicious for lymphoma, sarcoidosis, or infection. Mediastinal lymph node biopsy and an axillary lymph node biopsy taken 3 years and 2 months prior to presentation, respectively, did not demonstrate any diagnostic abnormalities; no evidence of lymphoma or granulomatous disease was present. A bone marrow biopsy specimen was only slightly hypercellular for age without clonal abnormalities on cytogenetic testing, which is consistent with CVID. Acid fast stains and culture for mycobacteria were negative.

Angiotensin-converting enzyme and serum calcium levels were normal at 35 (7–46) U/l and 9.1 (8.9–10.1) mg/dl, respectively. Her immunoglobulin levels were severely reduced [IgA 1 (50–400) mg/dl, IgM < 5 (50–300) mg/dl, and total IgG 183 (600–1,500) mg/dl].

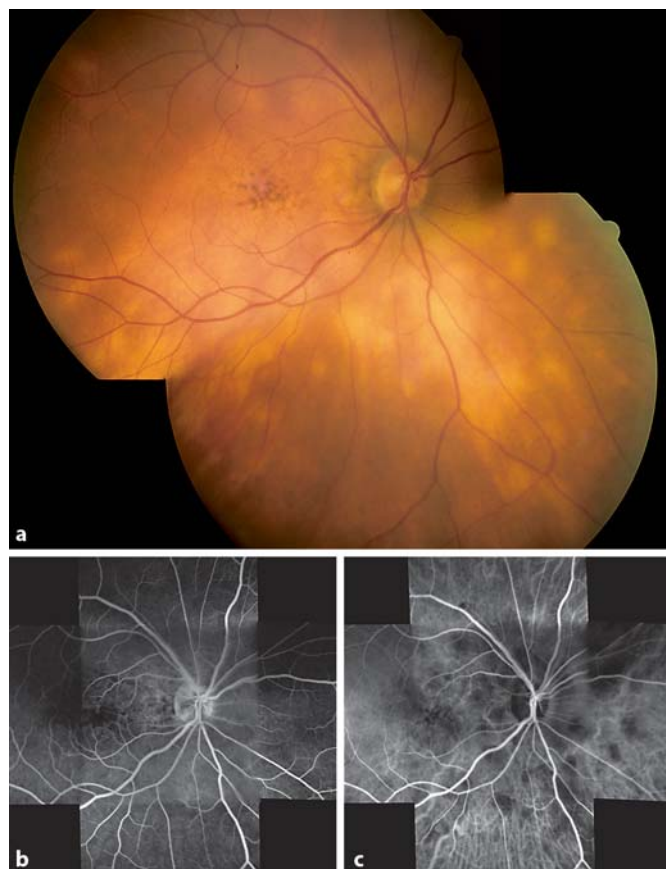


Fig. 1. **a** Fundus photo montage shows multiple hypopigmented placoid lesions outside of the macula and extending to the mid-periphery. Pigment alterations in the central macula are also seen. **b** Indocyanine green angiogram images (montages) showing multiple areas of hypofluorescence corresponding to the lesions seen on the fundus photographs. **c** Mid-phase (156 s) fluorescein angiogram.

Discussion

CVID is associated with a 5.4–10% incidence of non-caseating granulomas of lymphoid or solid organs [1] and an increased risk of malignant neoplasms, particularly non-Hodgkin lymphoma [2]. Sarcoidosis may also coexist with hypogammaglobulinemia [3].

Choroidal granulomas in sarcoidosis are well documented [4]. A Medline search revealed 1 case of CVID-associated choroidal granulomas in which sarcoidosis could not be ruled out as no lymph node biopsy was done despite elevated serum ACE level [5]. The diagnosis of choroidal granulomas was made without histopathologic evidence.

The patient presented in this report had normal serum ACE and calcium levels, normal biopsies of lymph nodes on two occasions and an essentially normal bone marrow, making a diagnosis of sarcoidosis or lymphoma unlikely. The absence of non-caseating granulomas in the lymph nodes also makes a diagnosis of tuberculosis very unlikely. Whether the observed diffuse placoid choriopathy is a result of CVID lymphoid hyperplasia, a local manifestation of CVID-associated granulomatous disease, or sarcoidosis is uncertain. However, it is impor-

tant to consider these relationships when evaluating CVID patients. The choroidal lesions may represent a similar process to the lymphadenopathy, rather than granulomatous disease or lymphoma. The long-term follow-up without deterioration suggests that these lesions may be of a benign nature. Of clinical importance, these findings may easily be misdiagnosed, and awareness of the ocular manifestations of CVID may prevent unnecessary and potentially harmful choroidal biopsy.

References

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