Clinical case

Unilateral orbital mass as an unusual presentation of IgG4-related disease

Masse orbitaire unilatérale : une présentation inhabituelle de la maladie à IgG4

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ABSTRACT

IgG4-related diseases are a recently recognized systemic syndrome characterized by mass-forming lesions, in mainly exocrine tissue, that consist of lymphoplasmacytic infiltrates and sclerosis, which may mimic malignant neoplasm due to clinical and imaging features. We report an unusual case of a 62-year-old woman who presented with a left orbital mass, which histologically revealed to be an IgG4-sclerosing disease.

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RéSUMé

La maladie à IgG4 est une entité de description récente caractérisée par la présence d’une ou plusieurs atteintes fibro-inflammatoires, surtout des glandes exocrines, pouvant mimer cliniquement et radiologiquement des tumeurs malignes. Nous rapportons un cas inhabituel d’une femme de 62 ans présentant une masse orbitaire gauche correspondant histologiquement à un tissu fibro-inflammatoire rentrant dans le cadre de maladie IgG4.

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1. Introduction

Few population-based studies of IgG4-related disease have been performed, and the epidemiology of the disease remains poorly described. The disorder is often identified incidentally through radiologic findings or unexpectedly in pathological specimens [1]. IgG4-related disease (IgG4-RD) is a systemic syndrome characterized by mass-forming lesions in mainly exocrine tissue that consist of lymphoplasmacytic infiltrates and sclerosis [2]. Although initially reported in some forms of pancreatitis, the disease also has been described in virtually every organ system: the biliary tree, salivary glands, peribital tissues, kidneys, lungs, lymph nodes, meninges, aorta, breast, prostate, thyroid, pericardium, and skin [3]. Furthermore, intracranial inflammatory pseudotumors associated with immunoglobulin G4-related disease can mimic multiple meningiomas [4,5]. Orbital IgG4-related disease (IgG4-RD) is rare, recently described but poorly understood which can occur in adults of any age. In this paper, we report an interesting case of a Tunisian woman who presented with IgG4-sclerosing disease isolated to the orbit and a review relevant literature on the subject.

2. Case report

A 62-year-old woman was admitted to our institution for a 6-month history of left proptosis. There was no previous medical history of any illness. Physical examination revealed a painless and irreducible exophthalmia with a well-limited tumor in the left upper eyelid. Preoperative MRI revealed an iso-intense orbital tumor of the left orbit on a T1-weighted image with low-density on a T2. There was no other evidence of head and neck or abdominal
involvement on MRI and CT scan (Fig. 1). Serology test results showed an elevated total IgG of 1916 mg/dL. Preoperative serum IgG4 was not performed in this case.

The mass was surgically removed. Macroscopically, the lesion was a 30 × 20 × 10 mm cm sized round solid mass with a smooth border and the cut surface was pale yellowish white and rubbery with streaks. Microscopically, the tumor was composed of a prominent lymphoplasmacytic infiltrate, with formation of numerous germinal centers within a fibrotic stroma. Immunohistochemistry showed numerous IgG4-positive plasma cells. The diagnosis of IgG4-RD was concluded. At 8 months after surgery, the patient showed no clinical signs of complications or recurrence (Fig. 2).

3. Discussion

Orbital tissues are affected by IgG4-RD. It was first observed that Mikulicz’s disease was correlated with IgG4-RD and later determined that IgG4-RD can occur in any ocular adnexal tissues [6]. The majority of entities were previously classified as chronic sclerosing sialadenitis, Mikulicz disease, and orbital pseudolymphoma, are now considered a part of the IgG4-RD spectrum [7]. Umehara et al. [8] proposed 3 criteria to diagnose IgG4-RD, which consist of the following:

- diffuse or localized swelling or masses in single or multiple organs;
- elevated serum IgG4 concentrations (≥ 135 mg/dL);
- histopathological examination shows (i) marked lymphocyte and plasmacyte infiltration and fibrosis and (ii) infiltration of IgG4-positive plasma cells with a ratio of IgG4/IgG positive cells > 40% and > 10 IgG4-positive plasma cells/HPF. The signs and symptoms of orbital IgG4-RD are chronic eye lid swelling and proptosis. Patients often have a history of allergic disease and elevated serum levels of IgG4 and IgE as well as hypergammaglobulinemia [6]. In cases of orbital IgG4-RD, 62% have bilateral lesions, 69% have lachrymal gland involvement, and 48% have bilateral lachrymal gland involvement [9,10].

The histopathological features bear striking similarities across organs, regardless of the site of disease. Elevated concentrations of IgG4 in tissue and serum are helpful in diagnosing IgG4-related disease, but neither is a specific diagnostic marker. Correlation with specific histopathological findings is essential, regardless of the serum IgG4 concentration, the number of IgG4-positive plasma cells in tissue, or the ratio of IgG4 to IgG in tissue [1].

Orbital IgG4-RD must be differentiated from: idiopathic orbital inflammation, marginal zone B-cell lymphoma, and reactive lymphoid hyperplasia with no IgG4-positive plasma cells. The exact role of IgG4 or IgG4-positive plasma cells in this disease has not yet been elucidated. The triggers and pathogenesis of IgG4-RD remain undefined [6]. Only some clinical features such as hypergammaglobulinemia and hypocomplementemia support the case for an autoimmunity nature [11].

Therapeutically, patients with IgG4-RD typically respond to immunosuppressive therapy with glucocorticoids [2,3], which decreases the size of the lesions, but relapse often occurs when systemic steroid therapy is discontinued [6]. A major determinant of treatment responsiveness is the degree of fibrosis within the affected organs. Untreated IgG4-related disease often progresses from lymphoplasmacytic inflammation to extensive fibrosis [1].

In conclusion, we reported here an interesting observation of orbital IgG4-RD presenting as a unilateral mass with no clinical signs of these diseases. The diagnosis was based on histopathological findings. Comprehensive understanding of the IgG4 molecule, the diverse facets of IgG4-related disease, and the response of this disease to treatment, may yield important insights into the immune system and other conditions now known to be associated with IgG4.

Disclosure of interest

The authors declare that they have no competing interest.

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