Atypical presentation of IgG₄-related disease as an isolated inferior orbital mass

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DESCRIPTION

A 24-year-old male patient presented to our specialty clinic with complaints of fullness in the inferior aspect of the left eye (figure 1A). The swelling was present from past 6 months and had increased to the present size. On palpation a postseptal mass was felt in the inferior orbit. The mass was painless in nature. On ocular examination, the visual acuity was 6/6 in both eyes. Pupillary reflexes were brisk. Fundus examination was normal, and no deficit of extraocular movements could be recorded. No other systemic abnormality was detected on general physical examination. The blood investigations revealed an increased erythrocyte sedimentation rate (ESR) (25 mm/hour). Chest X-ray and Mantoux test were done to rule out tuberculosis; however, these were found to be normal. Contrast-enhanced CT scan of chest and abdomen, serum antineutrophil cytoplasmic antibody (ANCA), antinuclear antibody (ANA), anti-ds DNA and rheumatoid factor levels were within normal limits. On ultrasonography of the left orbit, the echogenicity was consistent with that of a solid mass. A CT scan was done which revealed a non-specific mass in

the inferior orbit (figure 1B,C). The extraocular muscles were found to be normal. An excision biopsy was done through inferior orbitotomy which revealed a cellular lesion with multiple plasma cells (figure 1D). Immunohistochemistry was positive for IgG₄ (>40 plasma cells/hpf) (figure 1E). Serum IgG₄ levels were found to be raised as well (2.13 g/dL). A diagnosis of IgG₄-related disease (IgG₄-RD) was made and the patient was started on oral steroids (methylprednisolone) in a dose of 0.6 mg/kg/day for a period of 4 weeks followed by a gradual tapering over a period of 3 months. At 6-month follow-up, the patient remains in remission (figure 1F).

IgG₄-RD is an immune-mediated inflammatory condition which can affect various organs. The pathophysiology of this disease has not been completely understood. The diagnosis of IgG₄-RD requires a combination of clinical examination, imaging, histological and serological analyses. Orbit is a common site for involvement in IgG₄-RD, and it was the first extrapancreatic site to be reported.¹ Orbital IgG₄-RD (IgG₄ROD) is characterised by tumefactive orbital lesions showing polyclonal IgG₄ plasma cell rich lympho-plasmocytic infiltrate on histopathology with or without raised serum IgG₄ levels. It probably results from a dysregulated immune response to antigens that may involve several organs. In cases of IgG₄ROD, 62% have bilateral lesions, 69% have lacrimal gland involvement, out of which 48% have bilateral lacrimal gland involvement.² Within the orbit, the lacrimal gland is the most commonly affected structure, though it can affect any orbital structure. To the best of our knowledge, this is the first case of IgG₄ROD presenting as an isolated orbital mass. Extraorbital involvement occurs in 22%–71% cases of IgG₄ROD mostly to salivary glands and lymphnodes.¹ Some authors recommend 18-fluorodeoxyglucose (FDG) positron emission tomography (PET) to detect extraorbital involvement. There was no extraorbital involvement in current case. Glucocorticoids are the first line agents in the treatment of this disease entity. Rituximab (anti-CD20 monoclonal antibody) has emerged as a promising treatment option for IgG₄ROD.³

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Figure 1 (A) Clinical photograph showing inferior orbital swelling, (B, C) contrast-enhanced CT images in sagittal and coronal sections showing non-specific mass (arrow) in the inferior orbit closely abutting the inferior rectus. Histopathology revealed a cellular lesion composed of numerous plasma cells (D). Immunohistochemical stain for IgG₄ was strongly positive (>40 plasma cells/hpf) (E). (F) Clinical photograph at 6-month follow-up.

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Learning points

- ▶ Orbital IgG₄-related disease may present as an isolated orbital mass.
- ▶ A detailed work-up including histopathology is required to establish the diagnosis.
- ▶ Oral steroids are an effective treatment modality.

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