Incidental hypoglobus: primary amyloidosis of the superior rectus

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ABSTRACT

A 69-year-old man who presented with incidental hypoglobus was found to have an isolated superior rectus mass. Diagnosis of primary amyloidosis of superior rectus was made on incisional biopsy and negative systemic work-up. This is an unusual manifestation and site for amyloidosis and should be a differential of any extraocular muscle mass.

Keywords: extraocular mass, hypoglobus, orbit, primary amyloidosis, superior rectus muscle

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INTRODUCTION

Primary localised orbital amyloidosis is a rare condition, with isolated involvement of extraocular muscle being even less common. The clinical, radiological and histopathological findings of a case of localised orbital amyloidosis involving the superior rectus muscle are presented.

CASE REPORT

A 69-year-old Chinese man presented with a complaint of mild blurring of right eye vision for three months. Clinical examination showed reduced visual acuity of 6/24 and 6/18 in the right and left eyes, respectively, from moderate cataracts. Refraction did not reveal any significant astigmatism. He did not complain of diplopia and denied any history of trauma. An incidental asymptomatic finding of mild right hypoglobus with upper lid fullness was noted. Careful examination revealed a smooth, pinkish-white vascular mass overlying the right superior rectus. The posterior edge was not palpable (Fig. 1).

Restriction of right eye elevation and hypoglobus were demonstrated on Hess test. The right levator palpebrae superioris function and fundus examination were normal, and there was no proptosis on exophthalmometry. Non-ophthalmic physical examination was essentially normal. Computed tomography (CT) of the orbits performed with contrast enhancement showed a large well-defined mass in the superior orbit enveloping the superior muscle complex (Fig. 2). The superior rectus was thickened including the tendinous insertion, and enhanced (Fig. 3). The lacrimal gland and other recti muscles were normal.

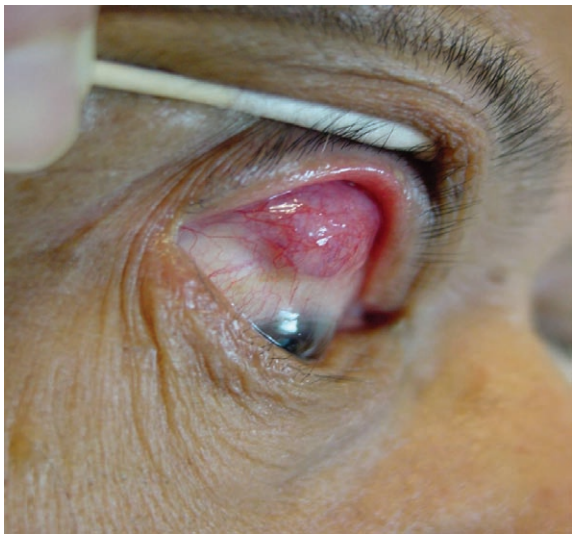

Fig. 1 Lateral photograph shows a smooth elevated pink vascular lobulated mass over the superior rectus.

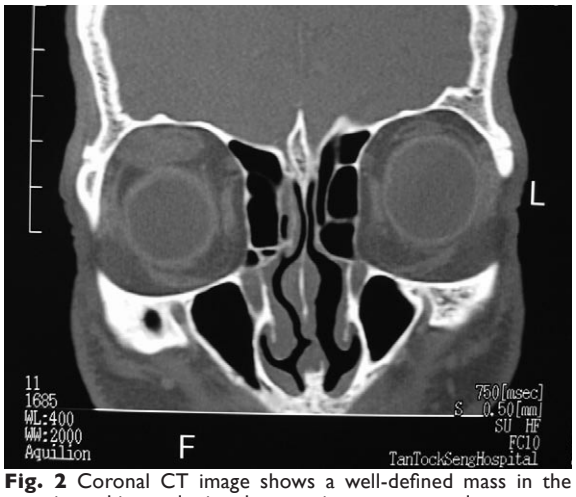

Fig. 2 Coronal CT image shows a well-defined mass in the superior orbit enveloping the superior rectus complex.

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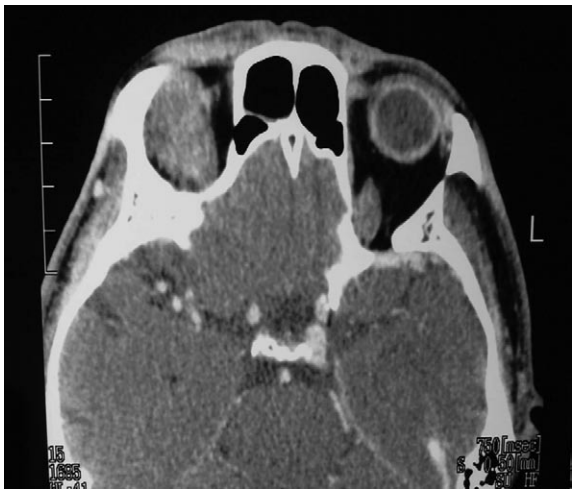

Fig. 3 Axial CT image of the orbits shows a lesion extending anteriorly to involve the tendon at the point of insertion.

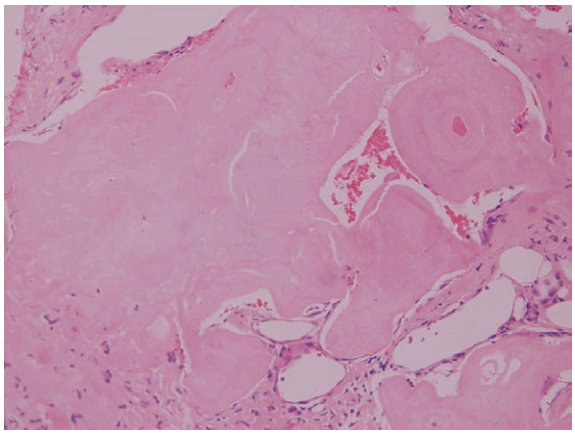

Fig. 4 Photomicrograph of the biopsy specimen shows nodules of amyloid (Haematoxylin & eosin stain, x20).

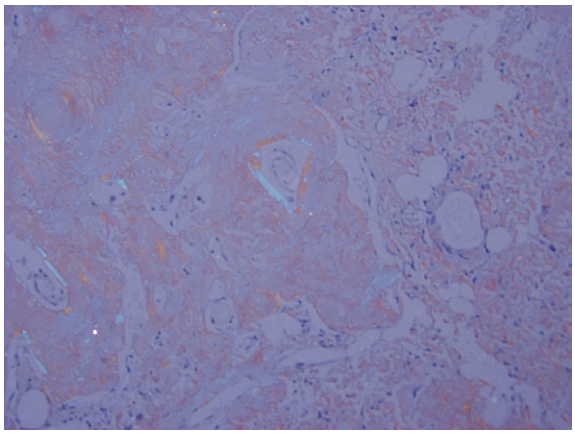

Fig. 5 Photomicrograph shows apple-green birefringence in polarised light (Congo red stain, x10).

In view of the clinical and radiological findings, an incisional biopsy was carried out. The mass was approached through the superior bulbar conjunctiva and revealed a smooth, soft, yellowish mass with no significant capsule. It was easily separated from the overlying conjunctiva and episclera, with minimal bleeding during biopsy. Histopathological examination demonstrated an amorphous mass with hyaline eosinophilic features on haematoxylin and eosin stain, and green birefringence with congo red-stain under polarised light consistent with amyloid (Figs. 4 & 5). Systemic review and investigations by the internist targeted at establishing secondary amyloidosis, were negative. In particular, the liver function tests, serum immunoglobulin levels, serum and urine electrophoresis for Bence-Jones proteins were all normal. Diagnosis of primary superior rectus amyloidosis was made.

On initial follow-up, the patient remained well with no systemic manifestations. However, the lesion gradually enlarged again after three years, resulting in upgaze restriction that required a repeat debulking surgery.

DISCUSSION

Amyloidosis is an idiopathic disease characterised by deposition of a heterogeneous proteinaceous extracellular material which may be found in any organ or tissue in the body. It is typically classified into primary, occurring de novo; or secondary to an underlying disease, which can be localised or systemic as characterised by multiple myeloma[1]. Localised amyloidosis is rare and its pathogenesis is largely unknown. It has been reported that only 4% of localised amyloidosis of the head and neck occur in the eye and orbits[2]. In the eye, amyloidosis more commonly affects the eyelids, conjunctiva and lacrimal gland, and is postulated to be a result of chronic inflammation such as trachoma[1-4]. Orbital involvement and infiltration of the extraocular muscles is rare and usually occurs in isolation. To date, there have been only six other reports of isolated extraocular muscle involvement[2,4-8].

Clinically, the most common presentation is a painless, palpable mass[2], as in our patient. Other presentations include proptosis, globe displacement, astigmatism and diplopia secondary to restricted motility[2]. Gross appearance of the lesion is often non-characteristic and ranges from a firm, pale, waxy amorphous mass to a pink, variegated lesion which may be easily mistaken for other ocular pathology such as lymphoma or metastatic tumours.

Radiological features are also non-characteristic. CT is the most commonly – used radiological investigation. Amyloid deposition in the recti has been noted to present with muscle enhancement and enlargement involving the tendinous insertions, features which were seen in our case. It may mould to adjacent orbital wall and globe and show adjacent

bony changes. Focal thinning and erosion, and hyperostotsis have been described[2,9]. On magnetic resonance imaging, amyloid may present as an area of heterogeneous hypodensity on T2-weighted images. On the fat-saturated contrast enhanced images, it appears as an area of marked homogeneous enhancement[9]. However, these findings are not specific to amyloid and may be confused with other diseases including granulomatous inflammation, lymphoproliferative disease, metastasis, and vascular disorders such as a cavernous haemangioma.

Definitive diagnosis is made through a biopsy of the lesion. Light microscopy of the specimen shows an eosinophilic extracellular substance that has an apple-green birefringence with polarised light and congo red stain. Histochemical differentiation can be made after pre-treatment with potassium permanganate ($KMnO_4$)[3]. Other investigative tools may include electron microscopy that demonstrates characteristic 70-100A° non-branching fibrils and β-pleated sheets on X-ray diffraction[3].

Management of amyloidosis involves investigation for an underlying systemic disease and especially paraproteinaemia. Tests may include chest radiographs, 2D-echocardiogram, serum and urine electrophoresis for Bence-Jones proteins, rheumatoid factor and anti-nuclear DNA. A rectal or abdominal fat pad biopsy is performed, when necessary, to confirm the diagnosis as these are positive for systemic amyloidosis in 80% of cases[7]. No effective treatment is available but extraocular muscle dysfunction with no significant enlargement may respond to strabismus surgery[7]. A large tumour causing troublesome visual symptoms or unsightly cosmesis may be best managed with debulking surgery[10].

The diagnosis of amyloidosis is an uncommon entity in the ophthalmological practice. It is often confused with other orbital pathologies and an incisional biopsy should always be performed to obtain histological diagnosis. This differential should be kept in mind for an atypical orbital lesion as there are potential serious systemic complications associated with it, although more often than not, it is isolated and benign.

REFERENCES

1. Howard GM. Amyloid tumours of the orbita. Br J Ophthalmol 1966; 50:421-5.
2. Okamato K, Ito J, Emura I, et al. Focal orbital amyloidosis presenting as rectus muscle enlargement. Am J Neuroradiol 1998; 19:1799-801.
3. Yakulis R, Dawson RR, Wang SE, Kennerdell JS. Fine needle aspiration diagnosis of orbital plasmacytoma with amyloidosis: a case report. Acta Cytol 1995; 39:104-10.
4. Holmstrom GE, Nyman KG. Primary orbital amyloidosis localised to an extraocular muscle. Br J Ophthalmol 1987; 71:32-3.
5. Erie JC, Garrity JA, Norman ME. Orbital amyloidosis involving the extraocular muscles. Arch Ophthalmol 1989; 107:1428-9.
6. Banerjee S, Bogman J, Reuser TT. Amyloid deposition in the extraocular muscles. Orbit 1999; 18:105-6.
7. Liesegang TJ. Amyloid infiltration of the levator palpebrae superioris muscle: case report. Ann Ophthalmol 1983; 15:610-3.
8. Ceviker N, Baykaner K, Akata F, Keskil S, Uluoglu Oe. Primary amyloidosis of an extraocular muscle. Neuroophthalmol 1997; 18:147-8.
9. Massry GG, Marrison W, Hornblass A. Clinical and computed tomography characteristics of amyloid tumour of the lacrimal gland. Ophthalmology 1996; 103:1233-6.
10. Patrinely JR. Koch DD. Surgical management of advanced ocular adnexal amyloidosis. Arch Ophthalmol 1992; 110:882-5.