CASE REPORT

Primary localised deep cutaneous amyloidosis of the eyelid

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Abstract

A 62-year-old lady presented with a six-month history of swelling of the left upper eyelid, resulting in mild mechanical ptosis. Clinical assessment suggested a provisional diagnosis of dermoid cyst. The lesion was excised and histology revealed nodular deposits of amorphous eosinophilic material surrounded by lymphocytes and plasma cells. Special histochemistry and immunoperoxidase stain results showed deposition of amyloid, non-AA type. The lesion recurred 6 months later.

Key words: Eye, eyelid, amyloidosis, primary localized amyloidosis

INTRODUCTION

Amyloidoses are a group of rare disorders characterised by extracellular deposition of several different types of proteins that share similar ultrastructural, immunofluorescent and histological features. Amyloidosis is classified into primary or secondary types, and is further subdivided into localised and systemic forms. Ocular amyloidoses may occur in the eyelids, conjunctiva, cornea, retina, extraocular muscles or in the vitreous. The eyelids are one of the most commonly involved sites in dermal and ocular amyloidosis. Cutaneous involvement of the eyelid is thought to be pathognomonic of primary systemic amyloidosis. Localised forms of amyloid deposits may also occur as a primary disease in the eye, or secondary to skin conditions like basal cell carcinoma, Bowen’s disease and seborrhoeic keratosis.

We report a case of primary localised deep cutaneous amyloidosis of the eyelid in the absence of systemic amyloidosis.

CASE REPORT

History

A 62-year-old Chinese female presented with swelling in the left upper eyelid of six months duration. There was no associated pain or redness of the eye. On examination of the left eye, a firm non-tender swelling measuring 15mm in largest dimension was noted in the outer one-third of the upper eyelid. The overlying skin was not attached to the swelling. There was mild mechanical ptosis. The lesion was excised and histology revealed nodular deposits of amorphous eosinophilic material surrounded by lymphocytes and plasma cells. Special histochemistry and immunoperoxidase stain results showed deposition of amyloid, non-AA type. The lesion recurred 6 months later.

Key words: Eye, eyelid, amyloidosis, primary localized amyloidosis

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Pathology
Grossly, both excision specimens showed brownish soft tissue (Fig. 1). Histology revealed deposits of amorphous eosinophilic material within fibrous tissue and blood vessel walls. These deposits were surrounded by some lymphocytes, plasma cells and foreign body-type multinucleated giant cells (Fig. 2). The Congo-red stain revealed salmon-pink deposits of amyloid, which showed apple-green birefringence on polarization. The deposits were potassium permanganate-resistant. Immunoperoxidase staining for Amyloid A (AA), as well as immunoglobulin light chains, was negative. A histological diagnosis of primary subcutaneous amyloidosis was made, based on the light microscopic findings and special stain results.

Further management
Following the tissue diagnosis in both instances, the patient was investigated for systemic amyloidosis. Full blood counts, erythrocyte sedimentation rate, serum electrophoresis, urine for Bence-Jones protein, liver function studies and renal function studies were performed and the results were within normal limits.

It was concluded that the amyloid deposits in this case was localised and primary in nature, with no evidence of systemic amyloidosis, pre-existing dermatological condition or plasma cell dyscrasia. The patient is currently well and remains on close follow-up.

DISCUSSION
Primary localised amyloidosis may be classified into three separate types - lichen amyloidosus, macular variant and tumefactive forms.\(^5\),\(^6\) The lesions of lichen\(^7\) and macular amyloidosis are mostly found on the lower limbs. The tumefactive form or nodular localised primary amyloidosis is uncommon and presents as single or multiple nodules in the extremities, trunk, genitals or face. Localised nodular amyloidosis has also been reported as primary focal lesions in the urinary tract,\(^8\),\(^9\) respiratory tract,\(^10\) colon,\(^11\) seminal vesicles\(^12\) and spine.\(^13\) Occasionally these lesions have been mistaken for malignant neoplasms and may cause significant morbidity.\(^14\) Localised nodular amyloidosis is not associated with any underlying dermatologic or systemic disease, but some cases appear to have a pre-existing inflammatory condition. Lin et al\(^15\) have documented 4 cases of localised nodular amyloidosis of the cornea secondary to trichiasis, of which 3 patients presented with progressively enlarging vascularized masses in the cornea.

In a study by Looi\(^16\) amongst Malaysian patients over a period of 5½ years, localised amyloidosis comprised 90.9% of all types of amyloidosis and of these 7.5% were primary localised cutaneous amyloidosis. These lesions were categorised into lichen and macular types based on clinicopathologic findings and it was noted that there were no cases of nodular amyloidosis. The findings of this study also indicated that the amyloid fibrils deposited in localised amyloidosis were potassium permanganate-resistant and of non-AA type – these findings concurred with that in our case report.

The pathogenesis of localised amyloidosis is thought to reflect either local production of
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Amyloid fibril precursors or the properties of the particular microenvironment.\textsuperscript{17} In localised amyloidosis the fibrils are of amyloid light chain (AL) type and may be associated with local proliferation of B-cells or plasma cells. As such, a B-cell or plasma cell dyscrasia must be excluded in AL amyloidosis.

The outcome of localised primary cutaneous amyloidosis is variable. Brownstein and Helwig\textsuperscript{5} reported progression of primary cutaneous amyloidosis to systemic amyloidosis in five of 10 patients, i.e. a progression rate of 50%. However, a more recent study by Woollons and Black\textsuperscript{6} showed that the progression to systemic amyloidosis occurred in only 7% of their patients.

Treatment\textsuperscript{2,17} of amyloidosis is tailored to each individual patient. In cases of tumefactive deposits of amyloidosis, surgical excision may be curative. However, cases with large amyloid masses may require debulking prior to surgical excision. Radiotherapy may be of use in reducing the size of the tumourous mass before surgery. Some patients have small lesions with no significant loss of function. In such cases conservative management of regular follow-up with close observation of tumour size may be of choice. However, local recurrence after surgical removal of cutaneous lesions has been reported.\textsuperscript{18} This may be attributed to conservative excision and persistent local disease.

In conclusion, primary localised cutaneous amyloidosis of the eye is an uncommon condition, which may be missed by the unwary. The presence of chronic inflammatory cells and foreign body-type multinucleated giant cells may lead to an erroneous diagnosis of a non-neoplastic inflammatory lesion. As extramedullary plasmacytoma of the orbit and eye is well-documented,\textsuperscript{19,20} it is important that this lesion is recognised so that an underlying treatable neoplastic disease can be excluded.

REFERENCES