Multifocal head and neck amyloidosis as a diagnostic clue of systemic lupus erythematosus (SLE)

A case report

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Abstract

Rationale: Amyloidosis accounts for 2% of head and neck tumors. Amyloidosis that develops in the head and neck region is localized amyloidosis. Multifocal amyloidosis in the head and neck region is extremely rare.

Patient concerns: The patient presented to the clinic of otolaryngology with nasal obstruction, anosmia and left neck mass for several months.

Diagnosis: A left nasopharynx tumor was revealed under nasopharyngeal scope. Eosinophilic, proteinaceous material was revealed under a pathology scope in the nasopharynx tissue and neck tumor. Congo red staining demonstrated pale congophilic amorphous material with apple-green birefringence under cross-polarized light, and multifocal amyloidosis was diagnosed. Amyloidosis secondary to systemic lupus erythematosus (SLE) was confirmed after a series of investigations.

Interventions: The patient underwent local excision for multifocal amyloidosis without following management. To control underlying SLE, the patient accepted steroid pulse therapy and immunosuppressants. The patient eventually achieved disease remission.

Outcomes: During the 6 months of follow-up in the outpatient department of otolaryngology and rheumatology, complications, recurrence of nasopharyngeal amyloidosis, and SLE flare-up were not observed.

Lessons: Head and neck amyloidosis involving the nasopharynx is a rare presentation of this disease. Head and neck multifocal amyloidosis should be taken as a hint of systemic disease. In head and neck amyloidosis, a comprehensive survey should be performed to clarify the underlying disease predisposing to amyloidosis and organ involvement.

Abbreviations: AA amyloidosis = reactive amyloidosis, AL amyloidosis = immunoglobulin light chain amyloidosis, SLE = systemic lupus erythematosus.

Keywords: amyloidosis, case report, head and neck, narrow-band image, nasopharynx, systemic lupus erythematosus

1. Introduction

Amyloidosis is a disease that results from extracellular deposition of insoluble misfolded fibrillar protein. Amyloidosis could be categorized into systemic or localized disease. Localized amyloidosis was defined as a single organ involvement of amyloid protein deposition and is rarely associated with systemic diseases. In contrast to localized amyloidosis, systemic amyloidosis involves more than 1 organ and multifocal lesions. Inflammatory diseases and some neoplastic diseases that produced misfolded fibrillar proteins, such as multiple myeloma and plasmacytic dyscrasia, should be concerned as the cause of systemic amyloidosis.\textsuperscript{[1,2]} However, the majority of amyloidosis discovered in the head and neck region was localized amyloidosis.\textsuperscript{[3-5]} Reviewing the previous literature, multifocal amyloidosis that developed in the head and neck region was extremely rare, and only 1 case associated with lymphoma was reported.\textsuperscript{[6]}

We reported a case of multifocal amyloidosis in the head and neck region resulting from SLE, and we would like to emphasize the importance of thoroughly investigating systemic disease in multifocal amyloidosis in the head and neck region.

The institutional review board of Taichung Tzu Chi Hospital approved this study. Informed written consent was obtained from the patient for publication of this case report and accompanying images

2. Case presentation

A 41-year-old female presented to the outpatient department of otolaryngology with nasal obstruction, loss of sense of smell and anterior neck mass for several months. An unmovable indurated
mass over the left submandibular area was noted. Ultrasound of the neck revealed a 3 × 2.2 cm round-shaped lymph node in the left level Ib. Left middle ear effusion was revealed by otoscope. Flexible nasopharyngoscopy demonstrated a bulging mass from the Rosenmuller fossa with yellowish smooth surface (Fig. 1A), which showed no angiodysplasia under narrow-band imaging (Fig. 1B). Further computed-tomography (Fig. 2) demonstrated a 2.2 × 1.5 × 2.2 cm enlarged lymph node in level Ib over left neck.

The pathology report of the excisional biopsy of left lymph node and nasopharynx revealed eosinophilic, proteinaceous material (Fig. 3A). Congo red staining demonstrated pale congophilic amorphous material with apple-green birefringence when viewed under high intensity, cross-polarized light (Fig. 3B), which were typical finding suggestive of amyloidosis. Multifocal amyloidosis was identified. Immunohistochemistry analysis was also performed and AA amyloidosis was confirmed by the presence of anti-AA antibodies.\(^7\)

Later, a complete blood count showed pancytopenia, a renal function test showed decreased estimated glomerular filtration rate and urine analysis showed proteinuria. Liver function test, chest radiograph and electrocardiogram all demonstrated normal results. Considering the clinical presentation and other laborato-

**Figure 1.** A. A bulging mass from the Rosenmuller fossa with a yellowish smooth surface. B. No angiodysplasia under narrow-band imaging.

**Figure 2.** A 2.2 × 1.5 × 2.2 cm enlarged lymph node at the level Ib of the left neck.

**Figure 3.** A. Eosinophilic, proteinaceous material in nasopharynx (40×). B. Congo red demonstrated pale congophilia with apple-green birefringence when viewed under high intensity, cross-polarized light (100×).
Amyloid precursors (AA, immunoglobulin light chains of type, anti-transthyretin). [1,2,7,8] Histochemical analysis with specific antibodies under high intensity, cross-polarized light and immunocytochemistry.

Clinical characteristics of rheumatic disease, such as rheumatoid arthritis, and ankylosing spondylitis. The gold standard of disease is rheumatoid arthritis, and ankylosing spondylitis. The larynx is the most common site of head and neck amyloidosis. In a 20-year retrospective study directed by Rudy et al, 2018, 22 out of 865 patients had amyloidosis involving the head and neck region (mostly larynx), and 4 of them had multifocal involvement. Only 1 case of multifocal head and neck amyloidosis was secondary to marginal zone lymphoma. [6] In this study, we presented a case of head and neck multifocal amyloidosis secondary to occult underlying disease. Although the incidence is rare, multifocal amyloidosis should be taken as a hint of underlying occult disease predisposing to multiple amyloid protein deposition. [14-16]

The head and neck region has numerous blood vessels and lymphatic ducts. Given that fibrils of amyloid protein would be deposited along the blood supply and lymphatic drainage, multifocal lesions should be of concern for disease involving the head and neck region. Although localized amyloidosis was identified initially, a thorough head and neck investigation was warranted.

Treatment of amyloidosis varies with the type and involvement of disease. Systemic amyloidosis carries worse prognosis than localized amyloidosis. [1,13] It is crucial to investigate and eliminate underlying disease in systemic amyloidosis as soon as possible. [16,37]

4. Conclusion

Head and neck multifocal amyloidosis should be taken as a hint of systemic disease. In head and neck amyloidosis, a comprehensive survey to clarify the underlying disease predisposing to amyloidosis and organ involvement is warranted.

Author contributions

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