

Haemoptysis & Purpuric Rash: A Rare Presentation of Amyloidosis.

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Introduction

Primary systemic amyloidosis (PSA) is a rare condition which often presents with multi-systemic involvement as a result of late presentation. Our case demonstrates the various mucocutaenous, and end-organ lesions that can manifest through amyloid fibril deposition in PSA. In addition, our case demonstrates how different modalities of diagnosis can vary and cloud the diagnosis, in particular the CT report and Serum Amyloid protein (SAP) scan result, which was not characteristic of amyloid deposition in the lung, even though the clinical picture suggested so.

Case Presentation

A 54 year-old gentleman presented to the respiratory clinic with a 2-3 year history of haemoptysis on exertion. He had developed progressive breathlessness and worsening exercise tolerance over the last 6 months, rendering him dependent on home oxygen therapy. He also complained of a recurring rash on his face and genitals (Image 2), impotence, a sore mouth and tongue, fatigue, myalgia, arthralgia and a recent development of dysphagia and diarrhoea.

On examination he had a diffuse, non-blanching, purpuric rash on his face and genitalia, as well as macroglossia, oral ulcers, sub-conjunctival haemorrhages and dystrophic nails.

On investigation our patient had high levels of serum lambda light chains and protein electrophoresis illustrated a dense band of IgG lambda protein (Table 1). The diagnosis was confirmed on a skin biopsy of the purpuric rash on his face (Image 1).

Discussion

Diagnosing primary systemic amyloidosis is difficult, and often delayed, due to the non-specific and varied symptoms at presentation. The disease itself is caused by amyloid deposition in the body's extracellular matrix and patients classically complain of the following: carpal tunnel syndrome, macroglossia, mucocutaenous lesions and oedema and examination often demonstrates hepatomegaly.^{[1],[2]}

Though diagnosing pulmonary amyloidosis appears to be rare, Smith *et al* documented that 88% of systemic amyloidosis patients had pulmonary involvement post-mortem^[8]. This indicates that pulmonary involvement is far more common than previously thought.

Conclusions

- Amyloidosis can present with a number of different clinical features. Early diagnosis is key to beginning curative therapy.
- Pulmonary involvement in amyloidosis is clinically rare but should be considered in patients presenting with haemoptysis, breathlessness and features of multi organ involvement. Investigations show characteristic restrictive pathology on pulmonary function testing.

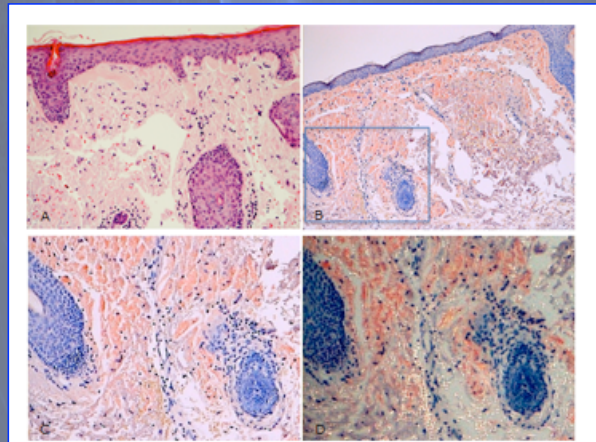


Image 1 (A) H&E staining from skin biopsy showing eosinophilic extracellular material within the papillary dermis. (B and C) Congo red staining of skin showing presence of amyloid as red amorphous extracellular material. (D) Same section under polarised light which shows red/green birefringent material confirming the presence of amyloid deposits (original magnifications (A) x 200 (B) x 100 (C&D) x 400

Table 1		
Investigation	Result	Normal Range
Electrophoresis	IgG 4.2g/L	5.3-16.5 g/L
IgG lambda paraprotein migrating within polyclonal immunoglobulin at fast/mig gamma.	IgA 0.37 g/l	0.8-4 g/L
	IgM 0.4g/L	0.5-2 g/L
Paraprotein concentration	2 g/L	
Free lambda light chains	930 mg/L	5.7-26.3 mg/L
Kappa/Lambda ratio	< 0.01	
Urinary protein creatinine	1.8 g/L	<0.21 g/L
Flow Cytometry		
An abnormal plasma cell population expresses CD38, CD138, CD56 and intracytoplasmic lambda light chains indicative of a plasma cell dyscrasia.		

Table 1 Results of further investigation with blood tests and protein electrophoresis

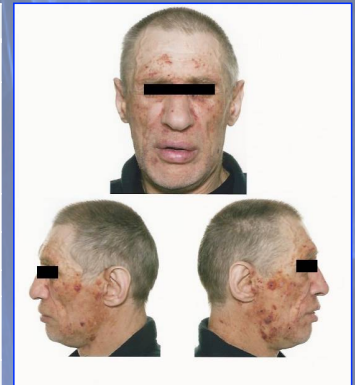


Image 2 Patient displaying purpuric rash

Five types of extravascular lung involvement have been described:

- Diffuse interstitial (or alveolar septal),
- nodular
- intra and extra-thoracic adenopathy,
- pleural
- (rarely) diaphragm disease^[3]

The Serum Amyloid Protein scan did not indicate pulmonary involvement in our patient however based on work by Smith *et al* it is highly likely that our patient's haemoptysis was secondary to amyloid deposition in the lungs. Given that our patient's haemoptysis has not recurred and that he no longer requires oxygen at home since beginning chemotherapy, it is more than likely that amyloid deposition was the cause of his haemoptysis.

References

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