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AL Amyloidosis: The Great Masquerade

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Introduction

- Rare, systemic disorder caused by misfolded immunoglobulin light chains from clonal plasma cells.
- Insoluble amyloid fibrils deposit in organs, leading to progressive dysfunction.
- Symptoms mimic other diseases like autoimmune or inflammatory diseases
- Commonly affected organs: Heart, kidneys, nerves, liver, and skin (Fig.1)

Case Presentation

Patient: 54-year-old male, cleaning supervisor

Initial symptoms:

- Duration: 4–6 months
- Swollen and painful hands
- Tight skin
- Numbness in radial aspect of L 4 fingers
- Muscle weakness
- Hoarseness & sicca symptoms
- Fatigue & weight loss of 10kg

Physical Examinations:

- Yellow waxy deposits on eyelids
- Diffuse fibrosed palmar fascia in hands (Fig. 2)
- Sausage-like swelling of all fingers
- Positive Prayer's sign
- Proximal weakness , MRC 4/5
- Schirmer's test: bone dry

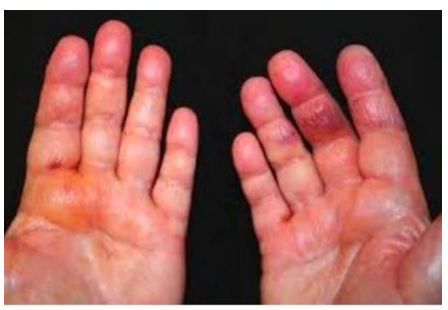


Fig 2: Fibrosed palmar fascia + tight skin

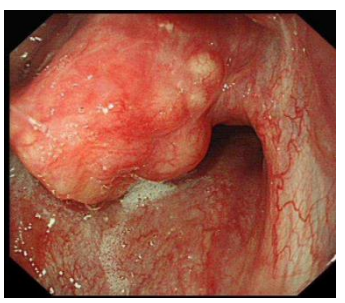


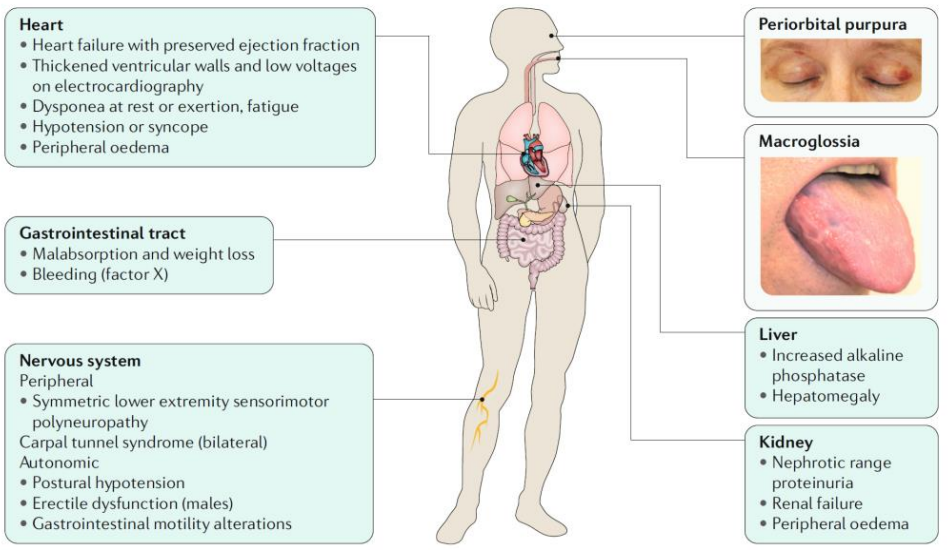
Fig 3: Amyloid deposit at epiglottis

Investigations:

- Nerve conduction study: bilateral severe carpal tunnel syndrome
- EMG: no feature of anterior horn cell disease
- Echo: Normal left ventricular systolic function, EF 60%. No regional wall motion abnormality seen. PASP 25mmHg
- CT neck, thorax, abdomen & pelvis: Small volume cervical and left supraclavicular lymph nodes.
- Creatinine 133; Creatine kinase: 40
- ESR 36, CRP 5.1
- Anti-nuclear ab (ELISA): 182%; Anti-Ro ab: positive
- Systemic sclerosis panel/ENA/ANCA/dsDNA: negative
- Paraneoplastic antibodies: negative
- OGD: Mild antral gastritis. Epiglottis looked deformed and hypertrophic. No ulcerations seen (Fig.3)

Initial suspicions: Scleroderma, Sjögren's, paraneoplastic syndrome

Clinical Presentation



Adapted from Bianchi et al. Hematol Oncol Clin N Am, 2020.

Fig 1: clinical presentation of AL amyloidosis.

Progress

8 months later:

- **Pancytopenia**
- **Cardiac & renal dysfunction**
- Blood – Hypogammaglobulinaemia
- Echo: **severe pulmonary hypertension, PASP 60mmHg, thickened valves**
- Renal biopsy: Glomerular mesangial proliferation, negative immunofluorescence and negative Congo red stain.

20 months later:

- **Macroglossia**
- Biopsies from palmar fascia: amyloid deposit
- Bone marrow biopsy: clonal plasma cells
- Dx: **AL amyloidosis + light-chain multiple myeloma**

Conclusion

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Diagnostic Challenges:

- Protean presentations → misleading
- Normal imaging and inconclusive biopsies delay diagnosis

Key learning:

- High clinical suspicion required for patients with **progressive unexplained systemic symptoms**
- Consider AL amyloidosis when:
  - ❑ Monoclonal gammopathy present
  - ❑ Multiple organs involved
- **Early diagnosis = better outcomes**

Reference

1. Bianchi G, Kumar S. Systemic Amyloidosis Due to Clonal Plasma Cell Diseases. Hematol Oncol Clin North Am. 2020 Dec;34(6):1009-1026