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PRIMARY SYSTEMIC AMYLOIDOSIS, PRESENTING AS PERIORBITAL PIGMENTATION

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A case of primary systemic amyloidosis with periorbital pigmentation is reported. She had mascular weakness, weight loss, paresthesia of hand, hoarseness, oedema, dyspnoea, macroglossia, dysphagia & purpura. There was extreme fragility of both involved or non-involved skin. Fat aspirate from abdominal subcutaneous tissue was positive for amyloid.

Key Words : Primary Systemic Amyloidosis, Periorbital Pigmentation

Introduction

Incidence of systemic amyloidosis is not available. In the studies based on routine post-mortem data in many general hospitals, prevalence of amyloid at autopsy was about 0.5%, that of Japan the figure is low (about 0.1%) and that when specialised hospitals were included or combined where a known genetic predisposition (such as Portugal and Isreal) existed the over all frequency is significantly greater. Ocular findings in primary generalised amyloidosis include (1) purpura of the eyelids, which can frequently be the presenting sign ; (2) bilateral, symmetrical, small amyloid papules of the skin of the eyelids; (3) ptosis; (4) opthalmagias or evidence of amyloid neuropathy affecting pupillar function or both and (5) subconjunctival haemorrhages.[1] Purpura of the eyelids following pinching (pinch purpura) and periorbital purpura following coughing, vomiting, the Vulsalva manoeuver, forced expiration spirometric testing and proctoscopy (post proctoscopic palpabral purpura PPP) are very characteristic.[2,3] Macular amyloidosis presenting as periocular hyperpigmentation have been reported.[4] We report a case of periorbital pigmentation in primary systemic amyloidosis.

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Case Report

A 60-year-old fair complexioned woman was admitted to our hospital because of dyspnoea specially on recumbency, pedal oedema, severe weakness confining her to bed and wheel-chair, weight loss, dysphagia and hoarseness of voice for the last one year. She had increasing symmetrical pigmentation of both the eyelids and periorbital regions leading to black eyes for the last 4 years (Fig. 1). For the same duration she was having recurrent purpura all over the body. Patient was a known case of bronchial asthma for the last 20 years. She also complained of paresthesia of right hand without loss of sensation. She noticed that her tongue was gradually enlarging for the last 1 year leading to gross macroglossia with superficial blisters, ulcers and purpura (Fig. 1). Tongue movement was very painful causing dysphagia for which she prefered to take liquid and semisolid food. Physical examination findings were; pulse 80/min, respiration 30/min and blood pressure 100/60 mm Hg. Pedal oedema was moderate. Liver was enlarged 3 cm. below the cost

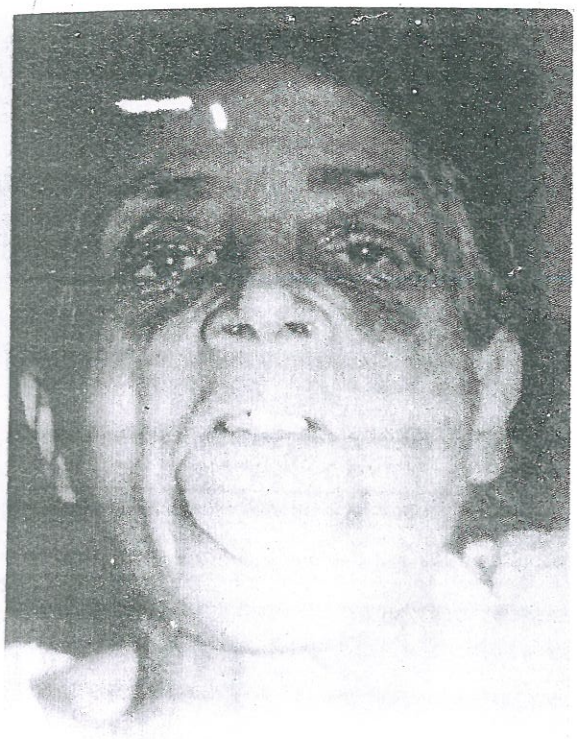

Fig. 1. Symmetrical pigmentation of both the eyelids and periorbital regions and gross macroglossia with superficial blisters, ulcers and purpuras.

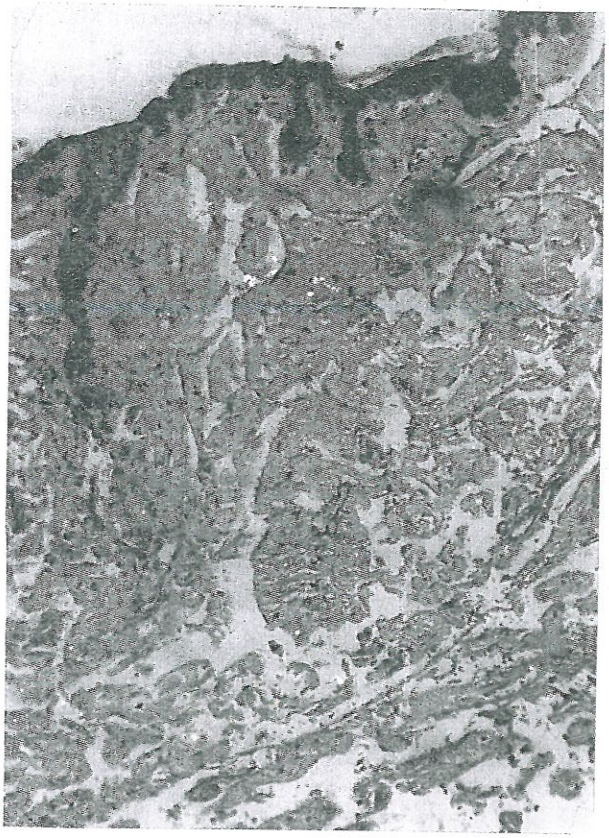

Fig. 2. Deeply pigmented epidermis and the entire dermis is homogeneously eosinophilic with deep fissures and infiltrate free (H&E, 10x10).

margin. Skin of eyelids and periorbital region was infiltrated, deeply pigmented with overlying papules and fragile. The noninvolved skin was also extremely fragile.The laboratory studies like routine blood count, liver function test, ECG, skeletal survey were normal. Bence Jones proteinuria was not found. Skin biopsy from upper eyelid showed deeply pigmented epidermis and the entire dermis was homogeneously eosinophilic with deep fissures and infiltrate free in H &E stain (Fig. 2). Biopsy from rectum and tongue were positive for amyloid but had intense infiltration with plasma cells and lymphocytes. The needle aspirate from subcutaneous tissue of abdomen showed eosin-stained fat cells. All tissues stained with Congo Red showed amyloid deposit.

Comments

Primary systemic amyloidosis is an uniformly fatal disease with median survival without myeloma is up to 14.7 months. Unusual longivity for 19 years have been reported with combined melphalan, prednisolone and colchicine treatment and is the longest reported in literature.[5] Because presenting symptoms are rather commonly nonspecific (including fatigue, weight loss, paresthesia, hoarseness, oedema, dyspnoea and lightheadedness or syncope secondary to orthostatic hypotension), there is often a delay (mean, 1-2 years) in reaching the diagnosis. So early diagnosis is essential to lengthen the survival period. Clinically evident mucocutaneous involvement in

primary systemic amyloidosis is an early pointer. Our case presented with periorbital pigmentation which to our knowledge has not been reported in primary systemic amyloidosis previously. Though a case of macular amyloidosis presented with periorbital pigmentation has been reported.[4] Another special feature of our case was extreme fragility of even noninvolved skin indicating presence of amyloid and this is in concurrence with the series where 16 of 34 biopsies from clinically noninvolved skin were positive for amyloid.[5] We also believe that aspirate of fat from abdominal subcutaneous tissue specimen is easy to obtain and very reliable (88% positivity) for diagnosis of systemic amyloidosis.

References

1. Golam A, Peyman, Donald R Sanders, et al. Amyloidosis. In : Principles and Practice of Ophthalmology, Ist Indian Edition. Philadelphia : W.B. Saunders, 1987 : 3rd Vol. 1650-1.
2. Stephen M B. Amyloid and amyloidosis. J Am Acad Dermatol 1988; 18 : 1-16.
3. Slagel A, et al. Postproctoscopic periorbital purpura. Arch Dermatol 1986; 122 : 463.
4. Van den Bergh WHHW, Starink TM. Macular amyloidosis, presenting as periocular hyperpigmentation. Clin Exp Dermatol 1983; 8 : 195-7.
5. Firtz DA, et al. Unusual longivity in primary Systemic amyloidosis, Am J Med 1989; 86 : 245.