

Systemic sclerosis and pheochromocytoma: A rare association

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Vol. 15, No. 1 (2011-01 - 2011-06)

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Abstract

Systemic Sclerosis is a multisystem disorder of unknown etiology. Peak age of onset is 30-50 year with male to female ratio of 3:1. Children represent fewer than 10% of all cases. Incidence of the disease is 0.1-14 per lakh general population. We report a 12 years old girl presenting with scleroderma and pheochromocytoma.

Key words : Systemic sclerosis , Raynaud's phenomenon, Hypertension, Pheochromocytoma
Accepted May 01 2010

Introduction

Systemic Sclerosis usually begins insidiously. The first symptoms are frequently Raynaud's phenomenon and puffy fingers. Raynaud's phenomenon is experienced in 95% of patients. Skin thickening is present in 100% of cases. The American college of Rheumatology has laid down the diagnostic criteria for systemic sclerosis [2]. Pheochromocytoma arises from chromaffin cells. The most common site of origin (90%) is the adrenal medulla. Ten percent occur in children, in whom they present most frequently between 6 and 14 years of age. They are found more often on the right side [3].

Case report

A 12 years old female child was brought with pigmentation and thickening of skin over extremities and face for 6-7 months, headache and palpitations for 4-5 months, tingling, numbness, pain, swelling in fingers and toes for 10 days. She was non-diabetic and did not complain of dysphagia or malabsorption. There was no loss of consciousness, cognitive decline or sphincter abnormalities. No family history of similar illness was present. She was lethargic, pale, undernourished but cooperative. Hyper-pigmentation was present over legs, feet, forearms, hands and face. Thickened patches of skin were present over feet and hands. Sloughing ulcers over toes were present (Fig.1). There was restriction in movement of fingers and toes due to hardening of skin. Recorded blood pressure was 180-240/100-130 mm Hg. Precordium was hyperdynamic. Apex beat was in 5th ICS and heaving in character. Palpable P2 and systolic thrill in pulmonary area was present. Parasternal heave was also present. On auscultation ejection systolic murmur of grade IV in pulmonary area and early diastolic murmur of grade III of functional aortic regurgitation in Erb's area were present. There were no signs of CHF and pulse rate was regular. Abdominal examination did not reveal any abnormality. Fundus examination showed hypertensive retinopathy of grade III. Patient was fully conscious and her SMR was of grade I.



Figure 1. Pigmentation and thickening of skin over ex-tremities



Figure 2. CT scan of abdomen showed adrenal mass on right side

Investigation revealed haemoglobin – 9.6 gm./dl, TLC 7400/cmm , DLC (P75L21 E4). ESR-55mm and platelets – 1.64 lacs/cmm. Urea/Creatinine (60.3/0.9 mg/dl), serum Na⁺/K⁺ (140/3.8 meq/lt.) serum alkaline phosphatase (36.3 IU/L) and ASO titre and liver function tests were normal. Urine routine microscopy showed proteinuria (+). She had a positive ANA but rheumatoid factor was nega-tive. Urinary VMA (47.9µg /gm of creatinine) was also positive. X-ray chest PA view showed cardiomegaly and pulmonary congestion. ECG showed left ventricular hy-pertrophy. Echo study showed aortic regurgitation, pul-monary hypertension and thickened myocardium.

Major criterion

Proximal scleroderma: typical sclerodermatous skin changes (tightness, thickening, and nonpitting induration, excluding localized forms of scleroderma) in-volving areas proximal to the metacarpophalangeal or metatarsophalangeal joints.

Minor criteria

- Sclerodactyly : sclerodermatous skin changes limited to digits
- Digital pitting scars resulting from digital ischaemia.
- Bibasilar pulmonary fibrosis not attributable to primary lung disease.

CT scan of abdomen showed adrenal mass on right side (5x5cm) (Fig.2). The patient was started on ACE inhibitors and β blockers but blood pressure was not controlled, so sodium nitroprusside was started but fluctuation of blood pressure still persisted. Salt restricted diet was advised. Ulcerations on the toes improved with conservative management. For Raynaud's phenomenon patient was advised to keep the limbs warm.

Diagnostic Criteria of Scleroderma

The diagnosis of scleroderma requires the presence one major criterion or two of the three minor criteria.

Discussion

Raynaud's phenomenon resulting from digital arterial spasm is often the earliest manifestation and may precede extensive skin and internal organ involvement by months or years. Raynaud's phenomenon, which is induced by exposure to cold, affects the fingers, toes and occasionally the ears and the tip of the nose. It has three stages: pallor, cyanosis and finally erythema. Two of three stages are considered sufficient for identifying this manifestation. Episodes can vary in duration from minutes to hours [1]. This patient had Raynaud's phenomenon. Raynaud's phenomenon is viewed as an important index characteristic for epidemiologic study of systemic sclerosis⁴.

Scleroderma means 'hard skin' and is the hallmark of the scleroderma spectrum disorders. The skin thickening of systemic sclerosis begins on the fingers and hands in nearly all cases. Superficial landmarks such as transverse digital skin creases are obscured and hair growth is sparse. The skin of the face and neck is usually next involved and is associated with an immobile and pinched facies. The skin changes may stay restricted to fingers, hands and face and may remain relatively mild. Some data suggest that untreated diffuse scleroderma is uniphasic with peaking of skin involvement in both extent and severity within 3 years.

Skin over the extremities, face and trunk may become darkly pigmented even without exposure to the sun. This patient showed hyperpigmentation and thickening of skin over hands and feet and also hyperpigmentation was present over face.

The sudden onset of accelerated to malignant hypertension, rapidly progressive renal insufficiency, hyperreninemia and evidence of microangiopathic hemolysis describes the syndrome of "scleroderma renal crisis"; [7,8]. The onset is most typically in cold weather months [8]. Profound decrease in the amount of haemoglobin may occur along with thrombocytopenia. Peripheral blood smears demonstrating fragmented RBCs can be key to an early diagnosis. Hematuria and proteinuria are followed by oliguria and renal failure. This patient had hypertension, hypertensive retinopathy and proteinuria. Pulmonary involvement has emerged as the leading cause of mortality and a principal source of morbidity. The pathologic processes in lung are vascular obliteration, fibrosis and inflammation. Progressive dyspnea on exertion, limited effort tolerance and a non productive cough are typical, whereas chest pain, pleuritic symptoms and increased sputum are less likely. Physical findings include fine early inspiratory crackling rales in the case of interstitial fibrotic disease or may reflect signs of pulmonary hypertension including an audible increased and palpable pulmonic component of the second heart sound, right ven-tricular gallops murmurs of pulmonary and tricuspid insufficiency, jugular venous distension, hepatjugular reflux and pedal oedema [9,10]. As a general rule, patients with diffuse scleroderma are at risk for progressive interstitial fibrotic lung disease. Individuals with limited systemic sclerosis may develop interstitial disease but they are also at a risk of progressive pulmonary hypertension in the absence of interstitial change, a complication most typical of long standing disease⁹.

The clinical features of pheochromocytoma result from excessive secretion of epinephrine and norepinephrine. All patients have hypertension at some time. Paroxysmal hypertension should particularly suggest pheochromocytoma as a diagnostic possibility. However, the hypertension in children is more often sustained rather than paroxysmal, in contrast to adults. Between attacks of hypertension, the patient may be free of symptoms. During attacks, the patient complains of headache, palpitations, abdominal pain and dizziness.

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