Systemic sclerosis and hereditary motor and sensory neuropathy: An uncommon combination

Sir,

Systemic sclerosis is one of the rare autoimmune connective tissue diseases with an incidence of 1/100,000.[1] Hereditary motor and sensory neuropathy is the most common inherited neuromuscular disease with an incidence of 10/100,000.[2] We report a patient with both systemic sclerosis and hereditary motor and sensory neuropathy which is as rare as 1/1,000,000 in the general population.

A 28-year-old male, harvesting machine driver, presented with tightening of the skin of limbs, face and claw hands. He gave a 2- to 3-year history of bluish discoloration of hands and feet on exposure to cold. He also had difficulty walking due to bilateral foot drop since childhood. There was a similar history of foot drop in his mother and paternal grandfather.

Cutaneous examination showed stretched, shiny, tight skin over the face [Figure 1], trunk and upper and lower limbs. Follicular hyperpigmented and perifollicular hypopigmented macules (salt and pepper pigmentation) were seen over the arms and front of the chest [Figure 2]. He had microstomia pinched nose [Figure 1], superciliary madarosis, symmetrical immobile total claw hands and wasting of the forearm muscles. Bilateral foot drop, sensory loss (callosities seen on the soles [Figure 3]), pes cavus and claw toes along with a high stepping gait were noted.

Dermoscopy showed a periungual pallor and a few thrombosed capillaries suggestive of avascularity. Distal hands and feet were cold with a bluish tinge. The modified Rodnan skin score was 32. Contrast-enhanced computed tomography of thorax was suggestive of early interstitial lung disease. Barium swallow showed Grade-1 esophageal reflux. The antinuclear antibody and anti-Scl 70 antibody were positive. Electrocardiogram and echocardiogram were normal. Nerve conduction study showed absent compound muscle action potential from both median and ulnar nerves and all the nerves of lower limbs with near-normal sensory nerve action potentials, suggestive of axonal injury to the affected nerves, that is, bilateral sensorimotor axonal polyneuropathy.

In view of superciliary madarosis, thickened peripheral nerves, claw hand and foot drop, a slit-skin smear was done to rule out Hansen's disease.

Sural nerve biopsy showed the normal structure of the nerve, without any inflammatory cells histopathologically. Skin biopsy was suggestive of scleroderma.

A diagnosis of systemic sclerosis with hereditary motor and sensory neuropathy (most probably Charcot-Marie-Tooth disease type 2) was hence made.

The patient was treated with cyclophosphamide pulse therapy, nifedipine for the frequent Raynaud's attacks, chest physiotherapy, occupational therapy for hands and feet, music and yoga therapy for his mental stress.

Systemic sclerosis is a chronic, multisystem connective tissue disease characterized by microangiopathy, immune activation and fibrosis of the skin and internal organs.[3] Systemic sclerosis can happen as a result of occupational exposure to silica, polyvinyl chloride trichloroethylene, organic solvents, pesticides, hair dyes and industrial fumes.[3]

In our patient, it might have been triggered by exposure to silica dust, him being a harvesting machine driver.

Skin sclerosis is a primary manifestation.[3] Raynaud's phenomenon is seen in 75% of the patients and ischemic digital ulcers by 50%.[3] Salt and pepper pigmentation is found to be a presenting feature in many in India.[4] Calcinosis cutis, leg ulcers, telangiectasia and sexual dysfunction are the other concerns.[3]

Nail fold dermoscopy progresses from isolated dilated loops to tortuous loops to total loss and avascularity in late stages.[5,6] Various autoimmune antibodies such as anti-serum anti-topoisomerase, anti-RNA polymerase III antibody and anti-centromere antibodies are associated.[7]

Charcot-Marie-Tooth disease is the most common hereditary motor and sensory neuropathy. Symptoms usually begin in early childhood but it can start at any age.[8] The initial symptom is foot drop and hammer toes. Wasting of muscles of the lower legs may give a "stork leg" or "inverted champagne

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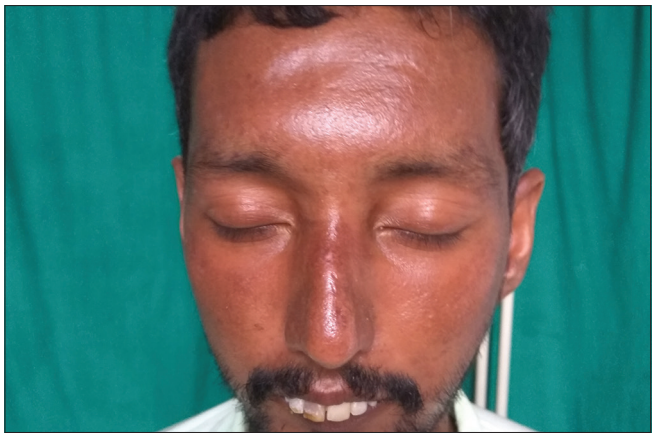

Figure 1: Stretched, shiny and tight skin of the face

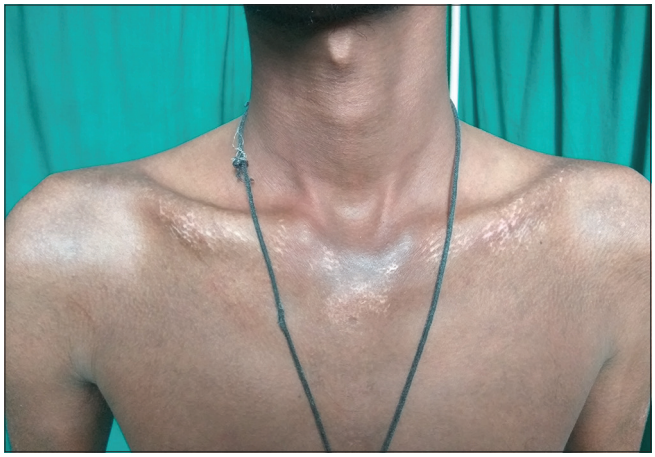

Figure 2: Salt and pepper pigmentation in front of the chest

bottle" appearance.[8] Weakness and sensory loss in the hands and forearms occur in many.[8,9]

Early- and late-onset forms occur with "on and off" painful spasmodic muscular contractions that can be disabling when the disease activates.[9] High-arched or flat-arched feet are classic associations of the disorder.[9] Overuse of an affected limb can activate symptoms including numbness, spasm and painful cramping.[8,9]

Electromyogram and nerve conduction study are used to know the extent of muscle and nerve damage.[8] Nerve conduction study is suggestive of whether it is demyelination or axonal degeneration.[1] Genetic studies are useful in detecting the mutant genes as around 90 genes are involved in the process of the disease.[9]

The coexistence of these two rare diseases in one person makes it rarer still and worth reporting. It could be a coincidence or an unexplained common mechanism could have triggered both the conditions.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

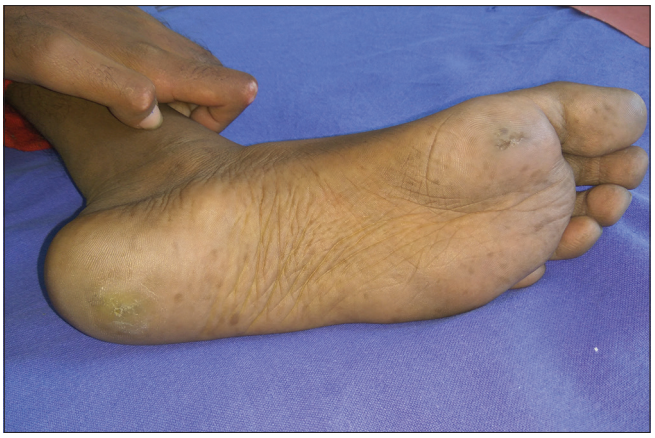

Figure 3: Loss of sensation over the sole seen as callosities

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Conflicts of interest

There are no conflicts of interest.

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