Total hemiatrophy as a rare presentation of linear scleroderma

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Abstract

Scleroderma rarely presents as total hemiatrophy of one side of the body. A 63-year-old patient is discussed with progressive facial hemiatrophy and with atrophy of skin, subcutaneous tissue, muscle and bone tissue of one side of the body, complicated by ocular and neurological manifestations.

Keywords: Scleroderma; Hemaatrophy; Osteoporosis

1. Introduction

Scleroderma is a connective tissue disease that is characterized by fibrosis, vascular obliterative disease and variable expressions of disordered immunity [1,2]. In rare cases scleroderma presents with progressive facial hemiatrophy (PFH) or Romberg's disease. We describe a 63-year-old female patient with PFH and complete atrophy of one side of the body.

2. Case report

At present our patient is a 63-year-old woman. Scleroderma started at the age of 15 years with progressive thickening of the skin, localized distally on her right leg, arm and hemi-thorax. From the age of 18 to 38 years the disease remained more or less stable. When the patient was 38 years old, she started complaining of loss of strength in her right hand. She had never used chemical agents, which can induce scleroderma-like conditions. On physical examination there was a progressive atrophy of muscles of the first and fifth fingers of her right hand with atrophy of the interossei muscles in the first four fingers. Small finger movements were impaired. The circumference of the right proximal and distal arm was 3 cm and of the right proximal and distal leg was 10 cm. Loss of sensibility of the fifth finger had occurred and there was a hypaesthesia in the rest of the right hand. The disease has remained stable until present.

Physical examination at present: a 63-year-old woman with extensive hemiatrophy of the right side of the body. Blood pressure was 100/70 mmHg, pulse rate 72/min. The central venous pressure was not elevated. On the face the right eye showed a small degree of ptosis and there also was anisocoria. In general the skin colour
was ivory and the right side of the thorax and the right arm also showed areas of brown hyperpigmentation. On macroscopic examination the affected skin seemed hairless and without sweat glands. There were subcutaneous calcifications of the right side of the body. The right arm and leg showed extensive muscle atrophy and the right hand showed sclerodactyly. The right breast was atrophic and the right nipple was smaller than the left one. Auscultation of the heart revealed a normal first and second heart sound. Over the lungs normal breath sounds were heard. Over the lungs normal breath sounds were heard. Liver and spleen were not enlarged. The anterior part of the leg showed a far extending ulcerative, dry-looking skin inflammation. The pulsations of the right posterior tibial and dorsal pedal artery were absent. The pulsations of the left and right femoral artery were normal. The right knee joint was deformed; there was limited mobility of the right glenohumeral and ankle joints. There was a contracture at the metacarpalphalangeal joints of the right hand. The quadriceps muscle and the distal leg showed atrophy. Neurological examination revealed loss of sensibility for pain and temperature in the right arm, leg and hemithorax. The tendon reflexes of the right arm and the Achilles tendon reflex were absent; the foot sole reflexes on both feet were plantar.

Laboratory findings: ESR 20 mm/h, Hb 8.9 mmol/l, thrombocytes 449/nl, leuKocytes 10.5 /nl with a normal differentiation, glucose 5.5 mmol/l, ALAT 13 U/l, LDH 192 U/l, AF 168 U/l, γGT 14 U/l, calcium 2.48 mmol/l, phosphate 0.82 mmol/l, albumin 38 g/l, cholesterol 4.5 mmol/l, triglycerides 1.47 mmol/l, magnesium 0.75 mmol/l, 25-(OH) vit D₃ 55 mmol/l, TT₄ 109 nmol/l, FT₄ index 105, T₃ uptake 0.96, TSH 0.66 mEq/l, cortisol 0.30 μmol/l, parathyroid hormone < 1.7 pmol/l. No specific antinuclear antibodies were detected.

The chest X-ray showed hyperinflation, an adhesive left sinus and small, old fibrotic changes in the right upper field. X-ray examination of the right arm and leg demonstrated subcutaneous calcinosis. X-ray examination of the distal leg showed a peristomal thickening and extensive calcification surrounding the tibia. X-ray examination of the right foot showed calcification specifically in the area of the naviculare, first cuneiform and medial sesamoidal bone beneath the first metatarsal. There was a hallux valgus and deformation of the interphalangeal joint of digit 1. Atrophy and shortening of the metatarsal 5; the basal digit 5 was missing. No signs of osteomyelitis were seen. The ankle-brachial index in our patient was lowered on the right (0.44 on the right and 1.00 on the left). Digital subtraction angiography confirmed a haemodynamic significant obstruction in the femoro-popliteal tract.

Electromyography (EMG) showed obvious signs of a peripheral neuropathy in the right arm and leg, while the arms were more affected than the legs. Treatment consisted of vitamin D, calcium and a biphosphonate for her hemi-osteoporosis and a femoral-popliteal bypass via the vascular surgeon. No skin biopsy was taken in view of the fact that the microvascular supply seemed impaired. The significant abnormalities on physical examination are shown in Figs. 1 and 2.

3. Discussion

This patient demonstrates a rare variant of scleroderma with total hemiatrophy. An extensive Medline-search revealed only a few cases described in the literature. Scleroderma or systemic sclerosis is a connective tissue disease characterized by fibrosis and vasculopathy [1,2]. It is classified in a localized and generalized form. Examples of a localized form are morphea, linear scleroderma and scleroderma en coup de sabre. Generalized scleroderma involves both the skin and internal organs. Progressive facial hemiatrophy or Romberg's disease was first described by Parry and Romberg (1825 and 1846). It is characterized by atrophic changes in muscle, bone and subcutaneous and adipose tissue [3]. Involvement of all one side of the body is extremely rare and only a few cases have been described in the literature [4–7]. The diagnosis is made from the clinical picture. In 1985 Kulo et al. described a 21-year-old female patient with atrophy limited to the left side of the body. This patient also developed Schönlein-Henoch nephritis and paroxysmal noc-
In 1992 Sakwaoka et al. described 5 cases of progressive facial hemiatrophy (PFH) [7]. In one case atrophy extended to the trunk and thigh. Our patient showed hemiatrophy of the cutaneous, subcutaneous, vascular and bone tissues. There was no clear involvement of the lungs, the digestive tract or the kidneys. The disease progressed only very slowly over the course of years. As discussed above, the few patients described in the literature vary considerably in description, although hemiatrophy of one side of the body is common to all patients. Ptosis and pupil abnormalities as described in this patient have been described in patients with hemifacial atrophy. Muchnick et al. [8] described 11 case histories of patients showing various ocular abnormalities such as eye lid abnormalities, and pupillary and iris disturbances. Our patient did not have involvement of the gastrointestinal tract, lungs, kidneys, heart or specific antinuclear antibodies, which is usually the case in patients with limited scleroderma [9,10]. Our patient had definite signs of peripheral neuropathy. Various neurological manifestations have been described in patients with progressive systemic sclerosis [11]: neuropathy, myelopathy and cerebrovascular disease.

Various disease-modifying drugs have been tried in the more generalized forms of scleroderma, such as penicillamine, corticosteroids, colchicine and other immunosuppressive agents...
with conflicting results. We did not use any of these drugs in our patient since the disease progressed only very slowly and there are no reports in the literature showing a beneficial effect of any of these agents in this particular disorder. In conclusion, we wish to emphasize that in patients with a localized form of scleroderma one should be careful not to miss abnormalities in vascular and bone tissue.

References