

## En coup de sabre accompanied by pachydermoperiostosis: a case report

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### ABSTRACT

*Scleroderma en coup de sabre, a variant of localized scleroderma is a disorder characterized by fibrosis of connective tissue. We report a 21-year-old female with scleroderma en coup de sabre accompanied by pachydermoperiostosis. She was born to consanguineous parents and her older sister also had pachydermoperiostosis characterized by clubbing of the digits, enlargement of distal parts of the extremities. The two disorders were diagnosed by clinical examination, histological and x-ray findings. In contrast to scleroderma, pachydermoperiostosis is a hypertrophic process characterized by periosteal proliferation of the tubular bones and hypertrophic skin changes. We discuss this interesting coexistence and review the literature.*

### Introduction

En coup de sabre is a type of linear scleroderma characterized by a linear band of atrophy in the skin that occurs in the frontal or frontoparietal scalp. This type is a very rare skin disorder and occurs most often during the first two decades of life. Various ocular and neurologic abnormalities have been reported in patients with en coup de sabre (1, 2). Here, we report a patient with linear scleroderma "en coup de sabre" (LSCS) accompanied by primary pachydermoperiostosis (PDP) that is a rare syndrome characterized by clubbed fingers and periostosis of the long bones.

### Case report

A 21-year-old female presented with linear alopecia and a depression on the frontal scalp and forehead. Her complaint has begun firstly as alopecia on the frontal scalp in August 2003. Then, linear alopecia and a depression developed prominently on her frontal scalp and forehead within one month. Although various medical treatments such as local potent steroid, colchicine and parenteral penicilline were prescribed, no response was achieved and she noted expansion of the lesion inferiorly on the forehead in one year. She has no family history of connective-tissue disease. A sclerotic, hypopigmented, linear plaque was present in

the middle of the forehead and frontal scalp on examination (Fig. 1). In addition, there was a linear alopecia in the area of the frontal scalp of the lesion. A complete blood count, urinalysis, erythrocyte sedimentation rate, eosinophil count, liver and thyroid function tests, growth hormone, fasting blood glucose, alkaline phosphatase, creatinine, urea, electrolytes, calcium, serum protein electrophoresis, rheumatoid factor, anticomplement and antinuclear antibodies were within normal ranges. IgM and IgG antibodies to *Borrelia burgdorferi* were negative. Histological examination of the biopsy taken from the sclerotic region on the frontal scalp revealed epidermal atrophy thick collagen bundles in the reticular dermis and superficial and deep perivascular infiltrate of monocytes (Fig. 2).

In addition to the findings, she had clubbing of the digits, enlargement of distal parts of the extremities, palmar and plantar hyperhidrosis for about 11 years (Fig. 3). There was thickening of the skin of the face, hands and feet, and prominent skin folds on her forehead. The rest of the examination was unremarkable. She was born to consanguineous parents and was the sixth of six siblings. Intrauterine growth retardation and delayed closure of the anterior fontanel were detected during the antenatal period and the early childhood, respectively. The patient's older sister had clubbing, thickening of the skin of her hands and feet, and enlargement of the distal ends of the extremities. Other members of the family had no abnormality. Electrocardiogram, chest x-ray and cranial magnetic resonance imaging were normal. Radiography of the long and flat bones was performed. X-rays showed subperiosteal new bone formation in the distal parts of the ulna, radius, tibia, fibula, metacarpals, metatarsals and phalanges bilaterally (Figs. 4 and 5).

On the basis of the clinical picture, histopathological and the radiographical findings, diagnosis of LSCS and PDP were made. She was treated with para-aminobenzoic acid potassium (4.5 gr per day) for scleroderma en coup de sabre (3). Skin softening developed and the lesion expansion stopped within 3

months. The treatment was stopped after six months.

### Discussion

LSCS is a subtype of localized scleroderma that usually occurs in the frontoparietal area with band-like sclerotic lesion and most often in the first two decades of life. Females are affected more frequently than males. LSCS involves superficial and deeper layers of the skin and it may be bilateral or extensive, causing facial hemiatrophy. Neurological (e.g. intracerebral calcification, epilepsy) and ophthalmological (e.g. enophthalmus, exophthalmus, iridocyclitis) involvement could be seen as rare complications of LSCS. Non-neurogenic ptosis and anomalies of the vertebral column associated with LSCS have been reported (4, 5).

As in other types of scleroderma, the etiology of LSCS is still unknown. Viral (e.g. *Cytomegalovirus*) or bacterial infection (e.g. *Borrelia*), exposures to environmental toxins, genetic factors, microchimerism and autoimmunity may play a role in this disease. The interactions between blood vessels, fibroblast activity, and immunological processes are very important in the pathogenesis of scleroderma. Fibroblasts isolated from scleroderma lesions and cultured *in vitro* are characterized by increased synthesis of collagen and other extracellular matrix proteins (6, 7).

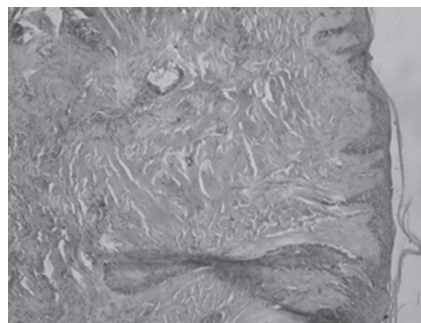
PDP is a rare hereditary disease characterized by osseous changes, skin changes, gastrointestinal changes and finger clubbing. Autosomal recessive and dominant inheritance has been suggested the inheritance pattern of PDP. In addition, it is stated that incomplete penetration may be responsible for the disease (8). Because our family has consanguineous parents, the condition appears to be autosomal recessive. Interestingly, our case does not have a brother and only the females are affected by PDP. The pathophysiologic mechanism of the disease is still unclear. Genetic influences, anomalies in fibroblast activity and alteration of peripheral blood flow have been suggested the etiological factors. It is stated that abnormalities of collagen synthesis in fibroblasts and vascular



**Fig. 1.** Sclerotic linear plaque in the middle of the forehead and frontal scalp.



**Fig. 2.** Broad collagen bundles that tend to parallel each other and epidermal atrophy (H&E, X10).



**Fig. 3.** Enlargement of distal parts of the extremities.

endothelial growth factor may play a role in the development of PDP (9, 10). Altered type I collagen expression and a correlation between the level of type I collagen mRNA and the transcriptional activity of the collagen gene in

PDP fibroblasts have been described by Padula *et al.* (11). They suggested that PDP could be accounted for by a trans-activated up-regulation of collagen transcription.

Matucci-Cerinic *et al.* (12) reviewed the pathogenetic pathways of scleroderma and PDP. These two diseases exhibit similar modifications in the microvascular system and connective tissue. In scleroderma, fibroblasts are activated and produce an impressive amount of collagen, leading eventually to tissue fibrosis. In PDP, the fibroblasts proliferate abnormally and produce large amounts of collagen and glycosaminoglycans in the dermis. Although the two diseases are both marked by the increased production of collagen, scleroderma results in cutaneous atrophy with epidermal thinning. On the other hand, PDP progresses to both epidermal and dermal hypertrophy. In conclusion, they hypothesized that the fibroblasts may be the determining factor in the course of two diseases but comparative studies should be performed about the behaviour of the fibroblasts in the two diseases.

In the present case, LSCS and PDP were observed together. We believe that, this coexistence may be remarka-



**Fig. 4.** Periosteal new bone formation on the hands.



**Fig. 5.** The tips of the fingers with irregularity of the distal part of terminal phalanges.

ble, because the pathogenesis of the two diseases has an alteration of fibroblast activity. The association of connective tissue diseases such as HLA-B27 positive sacroiliitis, rheumatoid arthritis and gout with PDP has previously been

described (9). To our knowledge, this is the first case with LSES that has atrophic process accompanied by PDP, a hypertrophic process. It is considered that occurrence of LSCS may be facilitated by PDP. We hope that this case

may contribute to the literature understanding of human fibrosis.

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