Chronic Scleroedema

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SUMMARY

Two patients with chronic scleroedema are presented. One patient had severe joint contractures. The literature on scleroedema is reviewed and the relationship of scleroedema to the stiff skin syndrome is discussed.

S. Afr. Med. J., **48**, 164, (1974).

The dermal deposition of mucopolysaccharide is found in genetic and acquired disease. Skin changes are described in the type 1 and type 2 mucopolysaccharidoses (Hunter and Hurler types) and in types 4 and 5 (Morquio and Scheie types). Skin changes in these patients are generally present over the hands, but may be more extensive. Histologically both epidermis and dermis are involved. Scleroedema, an acquired disease, is also characterised by excessive dermal mucopolysaccharides. The changes, which are said to spare the hands, may be localised or extensive.

The purpose of this report is to record 2 further cases with prolonged scleroedema and in whom the effect of the disease led to their attendance at an Arthritis Clinic for the evaluation of joint disease.

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Date received: 2 April 1973.

CASE REPORTS

Case 1

The patient, a 28-year-old woman, was first seen at the age of 11 years, after the abrupt onset of severe generalised stiffness of the skin. Examination at the time showed a thickened waxy skin over the chest, upper arms, neck and face, and to a lesser extent over the upper thighs. No treatment was prescribed, and during the ensuing years there was a general clearing of the skin. Re-attendance at the hospital 17 years later was necessitated by pain in the neck. Examination showed thickening of the skin over the upper arms and posterior aspect of the neck. The sternomastoids felt firm, but the skin of the anterior aspect of the neck appeared normal. The movements of the neck were normal, except

for a slight limitation of rotation and a marked reduction of extension. The rest of the physical examination was normal and X-ray films of the cervical spine revealed no intrinsic spine or joint disease. The limitation of rotation and extension of the neck appeared to result from involvement of the sternomastoid muscles.

Case 2

This patient, also a 28-year-old woman, was referred for evaluation of severe flexion deformities of the large joints of the upper and lower limbs. The deformities of her joints had developed during her lifetime and were associated with intermittent arthralgia in the knees and related to activity. Her family recalled that her limbs felt unduly firm from the age of 3 months. Despite increasing flexion contractures of her limbs, she had lived normally, although she had led a relatively sheltered existence as a result of her joint involvement. She complained of minor colour changes in her fingers on exposure to cold and of intermittent slight dysphagia.

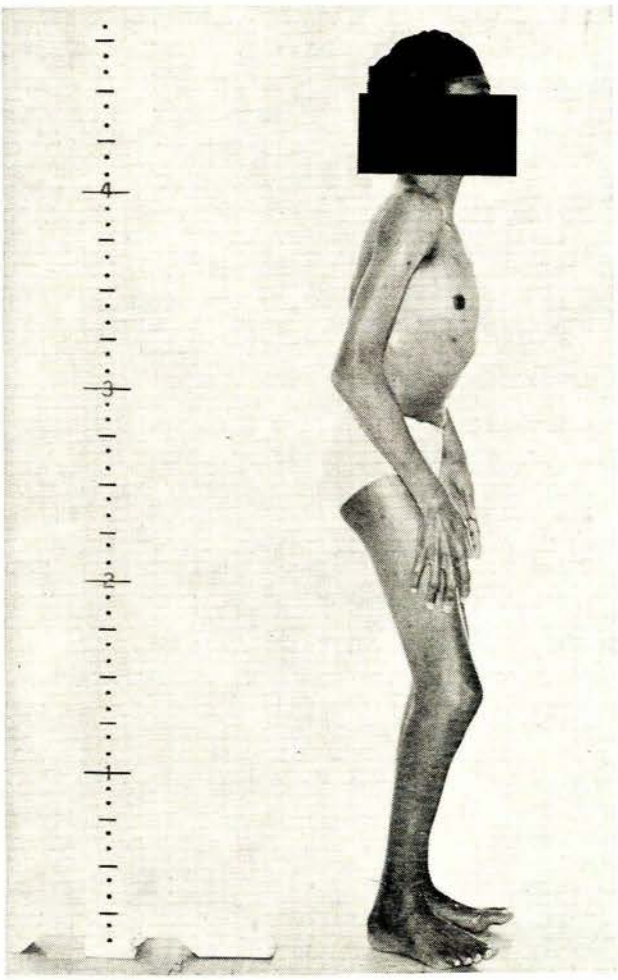

Fig. 1. Joint contractures of elbows, hips and knees (case 2).

Examination showed a thin, short woman with induration of the skin over the chest, abdomen, back and upper arms and thighs. The thickening of the skin was the result of involvement of the deep subcutaneous tissue. The induration over the anterior abdominal wall precluded an adequate examination of the abdomen. Chest movements were severely restricted. The patient was unable to stand erect, because of flexion deformities of the hips and knees (Fig. 1).

The cardiovascular and neurological examinations were normal. Haemoglobin was 6,5 g/100 ml, mean corpuscular haemoglobin 29,0%, serum iron 8 mg/100 ml, iron-binding capacity 430 mg/100 ml and saturation 2%. White cell count was 4 000/mm³ with a normal differential count. Bone marrow ratio to reactive marrow showed lack of iron. The sheep cell agglutination test and latex fixation test for rheumatoid factor and the antinuclear factor were negative. Creatinine clearance was 69 ml/min and the serum proteins, blood urea, and uric acid were normal. Wassermann reaction was negative and the electromyogram normal. The electrocardiogram inferior P vector was in keeping with pulmonary pathology. Pulmonary function tests showed a restrictive lung disease. Barium swallow and meal were normal, except for a small sliding hiatus hernia. X-ray films of hands, feet and spine showed mild, generalised osteoporosis, but no joint abnormality apart from the flexion deformities of the knees and elbows. The glucose tolerance test was normal, and there was no evidence of malabsorption. Skin and muscle biopsy specimens showed evidence of scleroedema (Fig. 2).

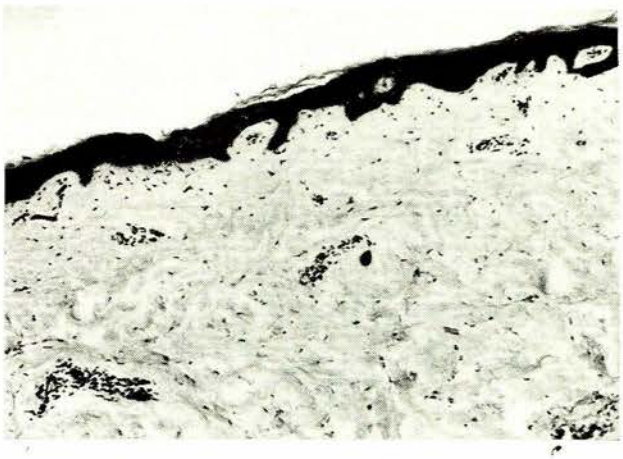

Fig. 2. Skin biopsy specimen. The dermis was infiltrated by mucopolysaccharide.

DISCUSSION

Despite the rarity of scleroedema, the disease has been well described in the German and English literature. The descriptions of the disease have been given by dermatologists mostly, and the major interest in the disease has centred on the infiltration of the dermis by mucopolysaccharides, which give the disease its clinical

and histological characteristics.[1-3] Three excellent reviews of the world literature have been reported and have served to broaden the earlier concepts of the disease.[4-6] Half the reported cases have occurred in childhood.

The earliest attempt to classify scleroedema appears to have been made by Janet *et al.*[7] who defined 2 groups: a benign childhood form, usually associated with a preceding infection and running a self-limiting course, and an adult form running a variable course. Subsequently, Robinow amplified the differences between these 2 forms,[5] and Graaf[8] reported that a study of cases collected at the Mayo Clinic led to the definition of 3 types, a self-limiting type associated with upper respiratory infection; an insidious type running a protracted course; and a third type resembling the second, but associated with diabetes mellitus. Several recent reports have emphasised the relationship of scleroedema with diabetes mellitus.[9,10]

Visceral involvement in scleroedema is well described[11-13] and muscle involvement appears to be most common. Cardiac involvement has been described.[14,15] The effects of the disease on joints have received scant attention. Joint immobility has been described in the early phase of the disease,[5] but there are very few descriptions of the protracted form of the disease. Schweitzer and Layman described a patient who had difficulty in rotating his neck.[16] Braitmann has reported a patient who complained of stiffness of the fingers and knees, but he did not detail any joint involvement.[17] One patient has been described who developed progressively impaired abduction of the shoulders.[18]

The 2 patients described here differ markedly. The first patient still has scleroedema involving the posterior neck and upper arms and has limitation of extension of the neck, with slight reduction in rotation of her neck after 17 years. The difficulty in this patient appears to be due to muscle involvement, particularly the sternomastoids. The second patient appears to have had scleroedema since infancy and has marked flexion deformities of her knees, hips and elbows, with total reduction of all movements of the spine and chest. There is no radiological evidence of joint disease in these patients and the severe joint deformities in the second patient are probably due to a combined effect of dermal and muscle involvement.

The *boutonnière* deformities of the fingers in the second case offer no easy explanation, and involvement of the sublimis muscle in the forearm is suggested, based on the explanation proposed by Casagrande[19] for similar deformities in rheumatoid arthritis in the absence of preceding proximal interphalangeal joint disease. No histological confirmation is available for the first patient, but in the second the histological pattern is consistent with a diagnosis of scleroedema and the special staining for mucopolysaccharides has shown that there is still residual mucopolysaccharide present in the dermis and muscle.

Many authors have discussed the differential diagnosis of scleroedema and have emphasised the importance of differentiating this from scleroderma, and dermatomyositis. Neither of these 2 patients shows evidence to support a diagnosis of scleroderma or dermatomyositis. Easterly and McKusick[20] have recently reported a few patients with a stiff skin syndrome and joint contracture which has histological similarities to symptoms in the patients with scleroedema. These authors considered this to be a new disease with a genetic basis, although the one patient described had no other family members involved, and was included because of the early onset of the disease. Congenital scleroedema has not been described, but Finkelstein[21] and Kutter[22] have each described scleroedema occurring in the early neonatal period. Our second patient is similar to the cases described by Easterly and McKusick.[20] The histological patterns of scleroedema and the stiff skin syndrome are identical, and they can only be separated by their clinical presentation. Under these circumstances it seems that in the sporadic case in a young child the diagnosis is largely a semantic problem. It is probable that both skin diseases, which are so much alike, are an expression of two different aetiological factors.

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