

## Progressive systemic sclerosis in childhood: A report of three cases

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### ABSTRACT

Systemic sclerosis is unusual in childhood. We describe three children who presented with diffuse hidebound skin associated with gastrointestinal and pulmonary abnormalities. Cardiac and renal dysfunctions, which are often encountered in these patients, were notably absent in our cases.

**KEY WORDS:** Childhood, Progressive systemic sclerosis

### INTRODUCTION

Scleroderma in childhood ranges from circumscribed and self-limited disease to diffuse skin involvement and multiple internal organ derangements with fatal outcome. Compared to adults, progressive systemic sclerosis is rare in young persons.<sup>1</sup> Only 1.5% of all cases occur under the age of 10 years, with females constituting at least two-thirds of the cases.<sup>2</sup> Here we present three children with sclerosis of the skin and associated pulmonary and gastrointestinal involvement.

### CASE REPORT

#### Case 1

A 9-year-old girl, born of a non-consanguineous marriage, presented with complaints of tightness of the skin which started over the hands, later extending to the forearms, face and trunk, with stiffness of the

hand joints since one year. She also complained of low grade fever, dysphagia and diffuse hair loss since six months.

On examination, the hands, forearms and trunk showed hidebound skin. The face revealed a pinched nose and microstomia with a positive Ingram's sign. Other findings were sclerodactyly with a positive Raynaud's phenomenon, stellate ulcers over the fingertips, and salt and pepper appearance of the skin over the forehead, extremities and back (Figures 1 and 2).

On investigation, anemia and raised ESR were detected. An X-ray of the chest was normal while that of the hands showed reduced periarticular bone density. Pulmonary function tests showed decreased vital capacity with restrictive lung disease. Barium studies revealed hampered peristalsis of the esophagus suggestive of dysmotility. The ANA, RA factor, VDRL and LE cell tests were normal. The skin biopsy was suggestive of

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**Figure 1: Sclerodactyly, thin lips and flattened lines of expression in case 1.**

scleroderma.

### Case 2

An 8-year-old boy, a product of a third degree consanguineous marriage, presented with restricted hand movements with tightness of the skin. A typical mask-like face, pinched nose, purse string mouth, sclerodactyly with Raynaud's phenomenon, and hidebound skin over the hands, forearms, arms and trunk were observed. Ulcers with a chalky white discharge suggestive of calcinosis cutis were noted on both elbow and ankle joints (Figure 3).

His pulmonary function tests were suggestive of a restrictive pattern with decreased  $FEV_1$  and vital capacity. Barium studies revealed sluggish peristalsis with dilated esophagus. The ANA, VDRL, RA factor and LE cell tests were negative. The skin biopsy showed collagenization of the dermis with high placed eccrine glands.

### Case 3

An 8-year-old girl, the first product of a non-consanguineous marriage, presented with characteristic features similar to the previous two patients. A skin biopsy was suggestive of scleroderma.



**Figure 2: Stellate fingertip ulcers in case 1.**



**Figure 3: Calcinosis cutis over elbow and back in case 2.**

Her pulmonary function tests revealed restrictive disease. Barium studies were not possible because the patient was uncooperative. The ECG showed tachycardia with right ventricular dominance. The ANA test was highly positive while the VDRL and RA tests were negative.

### DISCUSSION

Although resembling adult progressive systemic sclerosis (PSS), juvenile onset disease has a number of distinguishing features.<sup>3,4</sup> Similar to adult disease, it is characterized by Raynaud's phenomenon (90-95%), diffuse skin involvement and a microvasculopathy leading to progressive dysfunction of the esophagus, lungs, heart and kidney. All three patients presented with typical cutaneous features with two cases showing

calcinosis cutis.

Pulmonary involvement, though invariable in PSS, may remain asymptomatic or may present with dyspnea and cough. Although our patients were asymptomatic, they all showed a restrictive type of lung disease on pulmonary function tests.

Esophageal dysfunction occurs in most patients, although only half will complain of dysphagia.<sup>2</sup> One of our patients had dysphagia while barium studies revealed esophageal involvement in two patients.

Bodemer et al have reported that cardiac abnormalities like pericarditis, left or biventricular failure or arrhythmias are common in pediatric systemic sclerosis.<sup>5</sup> Only one of our patients showed tachycardia with right ventricular dominance secondary to pulmonary involvement.

Renal involvement is seen in 40-60% patients in the form of malignant hypertension, proteinuria, and azotemia. It generally occurs rapidly within the first three years of PSS and indicates a guarded prognosis.<sup>2</sup> None of our 3 patients have so far developed any clinical or laboratory abnormalities suggestive of renal dysfunction.

Antinuclear antibodies are reported in 70% to 90% of patients. Only one of our 3 patients showed ANA positivity.

All three patients and their families were counseled regarding the slow progression of this disorder and

supportive measures like protection from the cold, etc. As the beneficial effect of D-penicillamine is greater if it is started within the first three years, and considering the systemic involvement, it was started in all three patients in a dose of 3 mg/kg per day along with nifedipine. This dose can be increased every month up to a maximum of 10-15 mg/kg per day according to the response and tolerance to therapy.

The prognosis in childhood scleroderma depends on functional impairment and the course of visceral disease is difficult to predict.<sup>6</sup> However, Foeldvari et al have reported that most patients of juvenile systemic sclerosis show a favorable outcome and a significantly better survival rate as compared to their adult counterparts.<sup>7</sup> Thus the prefix 'progressive' is not uniformly applicable and should be avoided.

## REFERENCES

1. Lehman TJ. Systemic and localized scleroderma in childhood. *Curr Opin Rheumatol* 1996;8:576-9.
2. Singsen BH. Scleroderma in childhood. *Ped Clin N Am* 1986;33:1119-39.
3. Vancheeswaran R, Black CM, David J, Hassan N. Childhood onset scleroderma: Is it different from adult onset disease? *Arth Rheum* 1996;39:1041-9.
4. Blaszczy KM, Janninger CK, Jablonska S. Childhood scleroderma and its peculiarities. *Cutis* 1996;58:141-4,148-52.
5. Beemer C, Belon M, Hamel-Teillac D. Scleroderma in childhood: a retrospective study of 70 cases. *Ann Dermatol Venereol* 1999;126:691-4.
6. Foeldvari I. Scleroderma in childhood. *Curr Opin Rheumatol* 2002;14:699-703.
7. Foeldvari I, Zhavania M, Birdi N, Cuttica RJ. Favorable outcome in 135 children with juvenile systemic sclerosis: results of multinational survey. *Rheumatol* 2000;39:556-9.