# PROGERIA

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A 6-year-old male child developed progeria manifesting most of the typical changes described in progeria. The boy also had extensive sclerodermatous changes in the skin.

**Key words : Progeria, Sclerodermatous changes.**

Progeria (Hutchinson-Gilford syndrome) is characterised by retarded development and the onset of progressive senile degenerative changes in early childhood.[1] Scleroderma-like skin changes have been observed in a minority of progeria cases. The association of extensive and severe sclerodermatous changes in a case of this rare entity is being reported.

## Case Report

A 6-year-old male child had been having recurrent ulcerations on the skin for the last 4 years. The child was born after a normal delivery to non-consanguineous parents. He had delayed milestones. During the second year, the parents noticed hardening of the skin which gradually spread to the extremities and the face. Some of these areas broke down and resulted in ulcers with delayed healing. He began to lose scalp, body and eyebrow hair. He did not put on weight, though he had a good appetite, and had difficulty in using his limbs. Two sisters of the boy were normal. Physical examination revealed an extremely emaciated child, whose height of 91 cm (Normal—108.5 ± 7.15 cm) and weight of 8.5 Kg (Normal—16.3 ± 2.68 Kg) were low for his age. The face was bird-like in appearance with a thin, beaked nose. The facial skin was hyperpigmented and bound down. There were no hair on the scalp and eyebrows, lower eyelashes and body, except a few areas on the scalp and upper eye lashes. The eye balls appeared prominent and the child could not close his eyes because of scleroderma of the eyelids. The scalp skin was parchment-like and scalp veins were prominent. The left ear lobe was missing. The skin of neck, axillae, elbows, knees, parts of the buttocks and the genitalia showed fine wrinkling. The skin of the rest of the body was hyperpigmented, hard and bound down to deeper structures, with loss of subcutaneous and muscular tissue (Fig. 1). There were well defined areas of depigmentation on the left upper eye-lid, around the umbilicus and in the perianal region. There was a large,

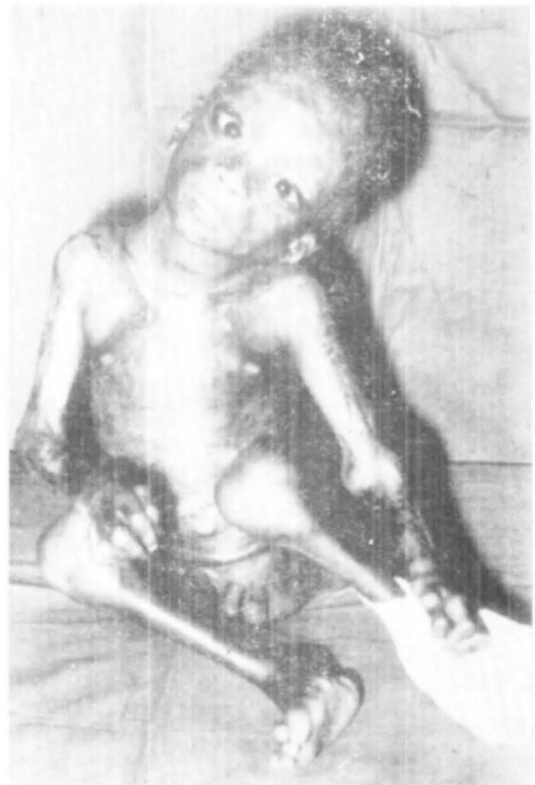

**Fig. 1.** Extensive areas of scleroderma and loss of subcutaneous tissues and muscles, alopecia of scalp and prominence of joints.

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non-healing ulcer, covered with slough on the dorsum of the left foot. The nails were normal. Sweating was noticed in the axillae but was absent in the sclerodermatous areas. His voice was thin and piping in quality. The teeth were carious and deciduous.

Elbows, knees and ankles were enlarged and prominent with fixed flexion deformities of elbows, knees and hips. The bones of the extremities were thin, small and tubular. The hands were clawed. The neck was flexed to the left. Frontal bossing with cranio-facial disproportion and slight micrognathia were also noticed. The anterior fontanelles were closed. The chest was barrel-shaped.

The liver was palpable 3 cm below the costal margin and was not tender. The heart and lungs were clinically normal. ECG was within normal limits. The child appeared mentally normal. Radiological survey of the skeleton revealed gracile long bones, generalised osteopenia, presence of unerupted teeth in the mandible, no widening of the periodontal space, ovoid configuration of the vertebrae (Fig. 2), bilateral coxa valga and hypoplasia of both clavicles.

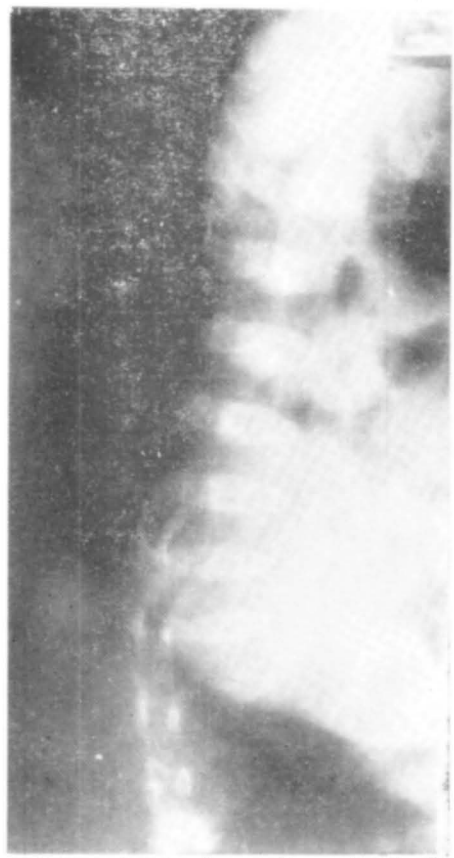

**Fig. 2** X-ray spine showing o oid vertebral bodies.

**Comments**

The classical features of progeria include short stature, diminished subcutaneous fat, low weight for the height, cranio-facial disproportion, recession of the mandible, prominent scalp veins, generalised alopecia, apparently prominent eyes, plucked bird appearance, pyriform thorax, short dystrophic clavicles, thin limbs with prominent stiff joints, horse-riding stance, coxa valga, delayed dentition, delayed sexual maturity, and wide based shuffling gait.[2] We observed almost all the features typical of this syndrome in our patient. In addition, the child also had extensive sclerodermatous changes, wrinkling of the skin in areas not involved by scleroderma, thin lips, absence of eyebrows, sparcity of eyelashes, and absence of the left earlobe.

Sclerodermatous changes were reported by Strinz et al. Moynahan,[3] who observed a morphoea type of scleroderma in his case, considered this early onset a striking and unique feature, and stated that in such cases of early appearance of scleroderma, the progeria picture would appear by the time the child was two years old. In Gabr et al's[4] cases of two sisters, the elder sister was found to have sclerodermatous changes. Marked induration and hyperpigmentation with poor muscle development of arms and legs have also been reported.[5] Scleroderma-like lesions were noticed in 4 out of less than 30 cases discovered till 1960.[3] It is also interesting to find vitiliginous areas in our patient. Depigmentation has not been observed in other cases of progeria.

## References

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