Progeria

A 12-year-old girl, weighing 9.0 kg (expected 39.5 kg) presented with growth failure, early onset of degenerative changes, alopecia, skeletal dysplasia and generalized atrophy of subcutaneous tissue since two years of age. The child had short stature (present length 94.0 cm; expected 149.0 cm) with a head circumference of 44.0 cm. Signs of premature ageing were present in form of generalized alopecia including loss of scalp hair, eyebrows and eyelashes; prominent scalp veins and a small triangular face with a relatively large cranial vault. The mandible was small with crowding of teeth (Fig. 1) and the nose was pinched. The skin was atrophic, inelastic, thin and dry with net like hyperpigmentation (Fig. 2). Nails were atrophic and brittle. Generalized wasting with cachectic appearance was evident.

Fig. 1. Note the typical old man appearance with loss of scalp hair, eyebrows and eyelashes, small mandible and crowded teeth.

Fig. 2. Skin over the dorsum of hands is atrophied, dried and hyperpigmented due to underlying atrophied subcutaneous tissue and fat.
pearance, generalized contractures, bony ankyloses and prominent joints were evident. The voice was high pitched and squeaky. Intelligence was normal. Radiological findings included delayed closure of cranial sutures, absence of clavicles, long slender limb bones with constriction at the upper end of radius (Fig. 3), acro-osteolysis of distal phalanges, and subcutaneous calcifications in lateral aspect of the pelvis (Fig. 4).

Fig. 3. X-ray elbow depicting constriction of upper end of radius.

Fig. 4. Subcutaneous calcification is most evident in radiographs of lateral aspects of pelvis.

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