**Introduction**

We report two patients with significant platyspondyly and distinctive peripheral radiographic findings. These children have a hitherto unreported complex bone dysplasia.

**Case reports**

**Patient 1**

During a diagnostic screening clinic in 1995 at a school for the physically disabled in Umtata, South Africa, we evaluated a boy aged 13 years with marked dwarfism. He had been born to young unrelated parents of Xhosa stock. He was an only child and there was no history of any skeletal disorder in the family. No other perinatal details were available.

On examination, he was of normal intellect but stunted, with a height of 124.5 cm (Fig. 1a,b). Head circumference was 54.5 cm (50th centile) and his weight was 30 kg. His face, teeth and palate were normal. He had a short neck, barrel chest and marked dorsal scoliosis. The contours of the shoulder girdles were abnormal with marked superior prominence of the acromion processes. There was restriction of movements, especially shoulder abduction. The elbows were bulky and lacked 20° of extension, as did the knees where marked valgus deformity was present. The fingers and toes were short and stubby. All other body systems were normal. In particular, liver and spleen were not palpable and no abnormality was evident in the eyes. For logistical reasons, it was not possible to undertake laboratory or histological investigations but chromosomal analysis revealed normal male karyotype.

Skeletal survey documented a generalised bone dysplasia (Fig. 1c–h). The shoulders showed dysplastic scapulae, which were irregular with areas of sclerosis and lucency and poorly defined glenohumeral margins. In the pelvis, similar, but more severe, changes mainly affected the acetabula, ischia and pubis. The spine was scoliotic with marked platyspondyly and slight irregularity of the vertebral end-plates. The clavicles were short and the ribs were narrow and oblique. There was severe epimetaphyseal involvement in the proximal femora. The remaining epiphyses and metaphyses were less affected, with changes being most marked at the knees, elbows, hands and feet. The carpal and tarsal bones were hypoplastic/dysplastic and the metacarpals and phalanges were short and broad. Pattern profile analysis confirmed shortening of all tubular bones of the hands, most marked in the middle phalanges (Fig. 2).

---

**Abstract**

Two patients with a unique generalised bone dysplasia demonstrating severe distinctive platyspondyly are reported. This group of crippling disorders defies metabolic and histological classification. The radiographic examination is, at present, the only practical method of documentation of these rare disorders.
Patient 2

This girl, aged 11 years 6 months, was seen at the Hospital of Polish Mother, Lodz for evaluation of platyspondyly and osteoporosis. Her bony abnormalities had been found incidentally at a regional hospital, where she had been hospitalised because of a respiratory infection. She was born at term, weighing 3100 g and length 52 cm. No abnormalities were detected at birth and the perinatal history was unremarkable. Her physical development was normal and, apart from the provision of orthopaedic shoes because of flat feet, she had been regarded as a normal child. She had not sustained any traumatic lesions and did not complain of joint pain. The parents were 24 and 35 years of age at the time of her birth. Mother was 158 cm in height and father 178 cm. Both were healthy, as were her five younger siblings.

Fig. 1a–h Patient 1 at 13 years of age. a, b Clinical appearance showing short stature due to short trunk, normal face, flexion deformities of large joints, genu valgum and short hands and feet. c Severe rectangular platyspondyly. d The iliac bones show reduced vertical height and the sciatic notches are narrow. The acetabular roofs are broad, horizontal and irregular. The pubic bones are thin and the symphysis pubis is wide. The right capital femoral epiphysis is small while the left is not ossified. The femoral necks are short. The lesser trochanters are prominent. There is marked flattening of L5. e Valgus deformity of the knees. The epiphyses are flattened and irregular in outline. There is minor metaphyseal involvement. f Distinctive half moon-like excavation of the distal humeri. g Thin, short anteriorly sloping ribs. h Short tubular bones with moderately severe epimetaphyseal involvement. The proximal phalanges show a unique metaphyseal peg indenting the epiphysis. Hypoplastic/dysplastic carpal bones. The distal radial and ulnar epiphyses are little affected.