Pseudoachondroplasia – Report of a South African family

Figure legends

Figure 1: Clinical photograph of the proband showing her disproportionate short stature, lack of facial dysmorphic features, marked genu varus and significant brachydactyly.

Figure 2: Clinical photograph of the proband’s father showing similar clinical phenotype, normal facies, genu valgus deformities and brachydactyly.

Figure 3: Spine X-rays (AP and lateral) of the proband: shows mild scoliosis, anterior beaking and tonguing of vertebral bodies.

Figure 4: Spine X-rays (AP and lateral) of proband’s father: shows marked scoliosis, degenerative changes, platyspondyly (probably secondary to compression fractures) and less beaking and tonguing of anterior vertebral bodies.

Figure 5: Proband’s knee X-rays (AP): shows significant metaphyseal irregularities (flaring and splaying) and small epiphyses.

Figure 6: Proband’s pelvic X-ray (AP): shows small capital femoral epiphyses, short femoral necks and irregular, flared femoral metaphyseal borders; poorly modelled acetabulae with irregular margins.

Figure 7: Proband’s X-ray hands (AP): shows significant brachydactyly, short metacarpals and phalanges with irregular metaphyses; small, irregular carpal bones.