A new form of spondyloperipheral dysplasia with facial dysmorphism, flattened vertebrae, hypoplastic pelvis, brachydactyly and soft tissue syndactyly

Abstract  We report the case of a 9-year-old Japanese boy with spondyloperipheral skeletal dysplasia associated with facial dysmorphism, pelvic abnormalities, and distinctive hands and feet. Radiographic manifestations included mild platyspondyly with posterior scalloping, small flared ilia with shallow acetabulae, mesomelic shortening of long bones, marked delay of carpal bone maturation, and brachydactyly with hypoplastic middle and terminal phalanges bilaterally in both hands and feet. There was bilateral soft tissue syndactyly of the 2nd and 3rd interdigital spaces of the hands, the 2nd interdigital space of the feet, with hypoplastic nails. The clinical and radiographic manifestations in this case appear to represent a unique type of skeletal dysplasia.

Introduction  We present a 9-year-old Japanese boy with facial dysmorphism, mental retardation, hypoplastic ilia, and characteristic hand and feet abnormalities associated with a generalized skeletal dysplasia with predominantly spondyloperipheral involvement. Spondyloperipheral dysplasia (SPD) is a rare skeletal dysplasia characterized by involvement of the vertebral bodies, hands, and feet. This entity has been considered by some as a particular type of spondyloepiphyseal dysplasia (SED) because it is often associated with generalized epiphyseal dysplasia. Since the first report of Kelly et al. [1], more than a dozen cases have been delineated [2–4]. To the best of our knowledge, however, the skeletal abnormalities observed in our case have not been reported previously in any other case of the SPD spectrum. The clinical and radiographic findings of this patient are depicted and discussed.

Case report  The patient, a 9-year-old Japanese boy, was the first child born to healthy, nonconsanguineous parents. His little brother was normal. The family history was unremarkable. He was delivered with a birth weight of 3,460 g, height of 51 cm at full term after an uncomplicated pregnancy. An Apgar score was 7/1 min. He was treated in an incubator for 4 days because of neonatal asphyxia. A chromosome analysis showed a normal male karyotype. No cutaneous or visceral abnormalities were found. Motor development is almost normal but he is mentally retarded (IQ 71).

He had a characteristic facial appearance, including a square face with an upward slant of the palpebral fissures, enlarged eyelids, strabismus, a broad nasal bridge, a thin upper lip, and a long philtrum (Fig. 1). His height at age 9 years was 125.5 cm (1 SD above the mean), while his arm span was disproportionately short (117 cm). Both hands and feet were markedly shortened and the nails were hypoplastic. There were soft tissue syndactilies of the 2nd and 3rd interdigital spaces of the hands and the 2nd interdigital space of the feet bilaterally (Fig. 2A,B). There were generalized joint contractures, including the hips, knees, ankles, elbows, and fingers. The external genitalia were normal.

The skull radiographs appeared normal. There was mild platyspondyly of the thoracic and lumbar vertebrae with posterior scalloping, and anterosuperior notching of the 4th lumbar vertebral
body (Fig.3). The pelvis was hypoplastic with small flared iliac wings and shallow dysplastic acetabula. Bilateral coxa valga was evident and the capital femoral epiphyses were slightly small for his age. The long bones in the lower extremities were overbroughted. There was mesomelic shortening of the long bones, especially in the lower extremities (Fig.4). The radiographs of the hands at age 4 years demonstrated that the metacarpals (mc) were short and slender with several pseudoeipiphyses (mc 2 + 5), the distal ends of the proximal and middle phalanges were pointed, and the terminal phalanges (2–5) were extremely hypoplastic (tulip shortening) (Fig.5A). The epiphyses were small for his chronological age, and only one small carpal center was ossified delineating dischonmonious ossification and severe bone age retardation. In the feet, the metatarsals were also slender with slightly cupped metaphyses. Ossification of the middle and terminal phalanges in the 2nd-5th digits was markedly delayed in contrast to the hands. Tarsal ossification appeared to be appropriate (Fig.5B). The terminal tufts in the hands and feet observed at age 4 years somewhat resembled those of acroosteolysis, but showed no changes of osteolysis subsequently in the recent films at 9 years of age (Fig.6A,B).

Discussion

We have delineated a unique type of skeletal dysplasia of facial dysmorphism, pelvic abnormalities in association with SPD and distinctive hands (and feet). SPD embraces a group of disorders with vertebral body abnormalities (platspondyly, end-plate irregularities) and brachydactyly [5]. The usual diagnostic criteria of SPD include a short stature, platyspondyly with inferior-superior notching (fish mouth vertebrae), broad hands with short fingers, markedly shortened distal phalanges, and short proximal and middle phalanges [4]. The radiographic findings in the vertebrae and hands of our case were consistent with this criteria, but he lacked the short stature. He not only had abnormalities of spondyloliperypheral involvement, but also a characteristic facial appearance, hypoplastic ilia with shallow acetabula, coxa valga, and syndactyly. The facial appearance of our case was different from that of the familial cases with SPD reported by Sorge et al. [4], which were the only previously reported SPD cases with facial anomalies. Also, the changes in the pelvis and hips observed in our case have not been noted in other cases of SPD.

Previously reported cases of SPD suggest an autosomal dominant inheritance pattern with considerable clinical variability [2, 4, 6]. The generalized epiphyseal abnormalities in some reported cases and an identification of a specific type II collagen gene mutation in a sporadic case of SPD support the view that at least some cases of SPD could be considered a special type of SED [7]. There were no obvious epiphyseal abnormalities in our case except for the hands and hips. The epiphyseal changes at the hips, which were mild, may be secondary to the poor formation of the acetabulum and abnormal weight bearing, and this may not really represent a primary epiphyseal dysplasia. The abnormalities of the hands and feet were most remarkable in our case. Many previously reported cases of SPD showed brachydactyly E-like digital changes, representing prominent shortening of the metacarpals.

Fig. 1 Characteristic square face with upward slant of the palpebral fissures, enlarged eyelids, strabismus, a broad nasal bridge, a thin upper lip, and a long philtrum.

Fig. 2 Brachydactyly with interdigital webbings of the 2nd and 3rd interdigital spaces in the hands (A), and the 2nd interdigital space in the feet (B), and hypoplastic nails.