CASE REPORTS

AN UNUSUAL OSSEOUS DYSTROPHY

BY

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The patient, J. C., whose physical and radiographical features are described in this paper, is a girl aged thirteen years. She has been under clinical and radiographic observation at The Royal Cripples Hospital since she was five years of age. She was born of normal parents who are unrelated; pregnancy was normal. There were no miscarriages, and the only other child (a younger boy, now aged ten) is normal. For the first two years of life the patient was thought to be normal, but after this time an obvious deformity of the spine developed for which treatment was sought from the late Mr. Naughton Dunn and since from Mr. F. G. Allan. She now shows a remarkable degree of dwarfing. At the age of five she was but 25 in. in height; this increased to 27 in. at the age of seven and to 28 in. at the age of thirteen in 1942. She weighs 38 lb. As will be seen from the photograph (fig. 1) she appears to have a very short neck and a marked spinal curvature, a severe scoliosis of the lower dorsal spine associated with a marked lumbar lordosis and a bilateral coxa vara. Abduction of the legs and arms is restricted—the arms cannot be raised above the shoulders. The hands and feet are broad and stubby. The relative lengths of the limbs

and trunk show no marked departure from the normal. There is no swelling of the peripheral joints and, apart from the limitations already indicated, movements of the limbs are free. She can run about and play with other children, but tires more quickly than they. She is a bright affectionate child and attends school with her brother. Owing to her repeated stays in hospital her education is backward, but her mother states that she is 'all there' and can be left with confidence in charge of the house and her brother, who is a boy of ten years of normal size and intellect (fig. 2). Her hair appears to be of normal sheen and coarseness.

FIG. 1.

FIG. 2.

There is no axillary or pubic hair, and no development of the breasts. No enlargement of the liver or spleen could be detected; the eyes appear to be clear and bright. The teeth are irregular and ill-developed and carious. In grotesque appearance she presents a superficial resemblance to the case of gargoylism (case 1, H. W., aged eighteen) described by Ellis, Sheldon and Capon (1936).

Radiographic examination

THE SKULL. No definite abnormality seen; all the sutures are shown. Normal sella. Sinuses are

heart being pushed over to the right. The ribs on the right show a marked compensatory curvature. No abnormality can be detected in the form or curvature of the vertebrae above the fourth dorsal (fig. 3 and 4). The clavicles are slender but otherwise normal.

THE UPPER EXTREMITY. The scapulae because of the curvature are elevated; they show poor development of the glenoid fossae.

Humerus.—The capital epiphyses, developed from three somewhat irregular ossific nuclei which have been compressed, are flattened and devoid of

FIG. 3.

FIG. 4.

well developed. All the teeth have erupted except the wisdoms which can all be seen in the radiographs.

SPINE. The vertebrae are of mature shape; there is no indication of vascular channels, epiphyses or delay in fusion of the elements. The lower part, which includes all the vertebrae from the ninth dorsal to the lower sacral forms a marked forward and regular curvature which includes the lumbosacral curvature. The dorsal vertebrae from the fourth to the ninth form a very marked convexity backward and to the left (a kyphoscoliosis) the

the normal ball-like articu'ar surface. The shafts are somewhat shorter than normal. The epiphyses for the lower end were late in developing and have not fused at the age of thirteen years.

Ulna and radius.—The nucleus for the head of the radius did not appear until the age of ten years. It is of greater density than the normal epiphyses and has not yet fused. The lower radial epiphysis was present at the age of five, together with nuclei for the os magnum and unciform. At ten years nuclei for the trapezium and trapezoid had been added as well as a smaller one for the left cuneiform.

At thirteen, nuclei for the scaphoid and semilunar have appeared; these are still rounded and present no evidence of their normal characters.

Hand.—All the epiphyses for the metacarpals and phalanges were laid down in single dense nuclei. At the age of five all nuclei were distinguishable with the exception of the terminal phalanges, base of first and head of the fifth metacarpals. At seven years the nucleus for the head of the fifth had appeared. At ten years a minute nucleus for the base of the first was recognizable; the epiphyses for all the proximal phalanges of the left fingers and the right ring and little fingers had fused; some had lost their unusual density and were now of the density of the diaphyses (fig. 5). At thirteen years all the epiphyses of the metacarpals and phalanges had fused and most had lost the unusual density which had formerly been a noticeable feature. No sign was seen of any epiphyses for the terminal phalanges.

The foot.—The tarsus, metatarsus and phalanges showed similar delay and radiographic appearances to those seen in the bones of the hand except that dense epiphyses for the terminal phalanges were present at the age of five years. In the case of the little toes the dense epiphyses for the terminal phalanges up to the age of thirteen were the only ossific representation of these bones. The opposing bony surfaces of the tarsus and metatarsus during development presented an unusually sharp cut angularity.

Differential diagnosis

GARGOYLISM. Ellis, Sheldon and Capon (1936) gave this name to patients having the syndrome 'a peculiar type of osseous dystrophy, congenital clouding of the cornea, enlargement of the liver and spleen and mental deficiency.' They chose the

FIG. 5.

(Note difference in ossification of terminal phalanges of toes.)

THE LOWER EXTREMITY. At the age of ten all the bones of the lower extremity showed marked osteoporosis.

Pelvis.—Regular in shape. The three elements were still distinct.

Femora.—At five years the capital epiphysis was ovoid in shape. The neck showed fragmentation and marked displacement as in severe cervical coxa vara. The epiphysis for the great trochanter was present (fig. 6). At thirteen years all the epiphyses and the fragments of the neck had soundly consolidated in a coxa vara position. The lower epiphyses and those of the upper and lower ends of the tibiae and fibulae were present at the age of five years (with the exception of the lower epiphyses of the tibia and fibula) and had fused at the age of thirteen (fig. 7). The diaphyseal extremities were rather irregular in outline and coarse in cancellous texture.

name 'on account of the large head, grotesque, inhuman facies and deformed limbs which suggest the appearance of gargoyles and render the patients closely similar to one another and the condition easily recognizable.' Certainly this patient bears a striking resemblance to their case 1 (H. W. female, aged eighteen years), but the radiographic characters and certain clinical features are entirely different. Apart from an osseous dystrophy this patient does not present any of the other features of the syndrome. The osseous dystrophy also is different. In gargoylism the vertebral bodies in the dorso-lumbar area tend to show a grooved anterior surface and one or more may be tongued, and at this level a kyphos is produced by backward displacement of a body slightly smaller than the others. There is a bilateral coxa valgus deformity, an enlargement of the sella, and delay in fusion of the epiphyses, but no unusual density and absence of the epiphyses.

CHONDRO-OSTEO-DYSTROPHY. The original case of this dystrophy, the clinical and radiographic features of which was shown at the Royal Society of Medicine in 1928 (Brailsford, 1929), when the patient was three years of age, presents some features which resemble the present case. It is interesting to know that these two children lived near to one another and played together for several years, but were unrelated. The patient W. T. B. presented radiographic appearances of the spine which resembled an exaggerated form of those seen

FIG. 6.

in gargoylism; he had a prominent kyphosis but he was a bright intelligent good-looking child. Multiple ossific nuclei developed in all his epiphyses, his joints were all swollen. He had no unusual density of his epiphyses and these appeared and fused at normal times. He was kept under periodic radiographic examination until he died aged fifteen in October, 1940. From the age of four years his musculature degenerated. At the age of thirteen years he was 2 ft. 11 in. in height, having gained but 2 in. since he was three years nine months. During the last three years of his life his limbs were flaccid. He was pallid, and periodically he had 'turns' during which he became blue and had profuse sweating from a cold skin. Radiographs in 1939 showed no increase in the length of the limbs or trunk. He then measured 2 ft. 9 in. The long bones were thicker, but the pressure deformities at the ends were more marked. The nuclei for the epiphyses at the knees had fused, but most of the other large epiphyses were fragmented. The vertebrae were compressed and irregular in outline. All the

FIG. 7.

bones showed osteoporosis and none showed normal cancellous structure. The joints were all swollen and the interosseous spaces increased. His appearance, distinct from that shown in my original photographs, was finally that of a most grotesque dwarf.

Summary

An osseous dystrophy is described, characterized by dense epiphyses which fuse early though development of other ossific centres is markedly delayed. Grotesque dwarfing with apparently normal intelligence is associated with severe spinal curvature and bilateral coxa vara.

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