A CASE OF DYSPLASIA EPIPHYSIALIS PUNCTATA

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A Coloured male infant aged 5 days presented at the Karl Bremer Hospital with some deformity of the skeletal system. The patient was referred to the X-ray Diagnostic Department on 22 June 1965 with a tentative clinical diagnosis of achondroplasia. This diagnosis was suggested because of the stunted and thickened appearance of the upper arms and the apparent large size of the skull. Some bluntness of the fingers was present and the flexed hands in the accoucheur position was a characteristic feature. The blood picture showed no changes of any pathological significance, and the Wassermann reaction was negative.

X-ray examination showed:

1. A skull of slightly larger than normal size with all the sutures and the anterior fontanelle completely closed.

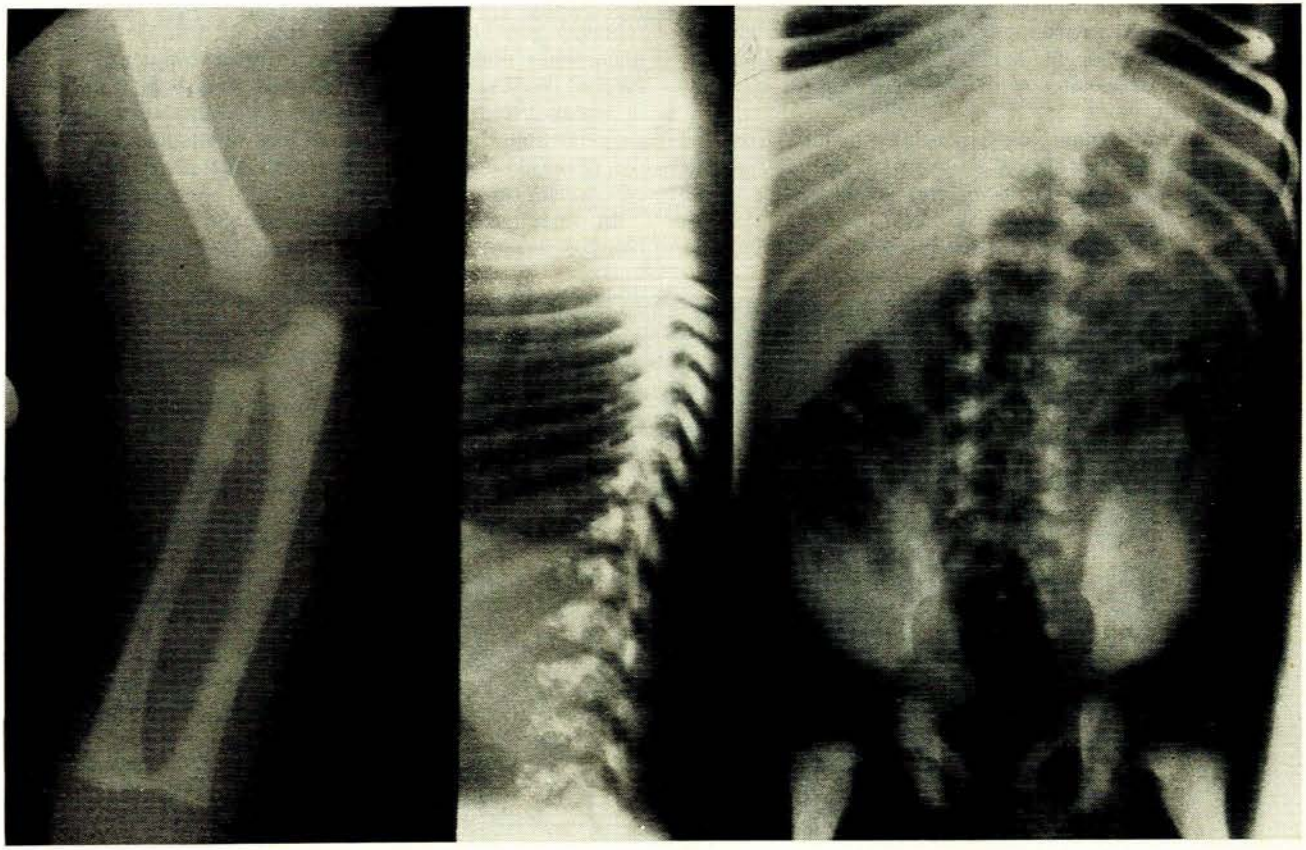

Fig. 1 *Fig. 2* *Fig. 3*

Fig. 1. Shortened and bevelled humerus. *Fig. 2.* Punctate ossification in the vertebral bodies. *Fig. 3.* Stippled and somewhat eccentric epiphysis of greater trochanter and apparent subluxation of hips. Pelvis normal.

No signs of increased intracranial pressure are observed and the pituitary fossa appears normal in size and shape.

2. No pathological changes in the lungs, and a heart shadow of normal appearance.

3. Short and thickened humeral shafts which are curiously bevelled at their proximal metaphyseal ends (Fig. 1). Peculiar punctate stippling of the epiphyses of the upper tuberosity of the humerus is present and there is an apparent subluxation of the shoulder joints. These affected epiphyses appear curiously eccentric.

4. 'Paint-brush flicking' in the vertebral bodies which are ossifying from numerous centres (Fig. 2). Adjacently one or two calcyeal spots in the intervertebral discs are observed.

5. Pelvic bony structures normal, but the hip joints appear subluxated and distinct separate centres of ossification are present in the trochanteric regions. These stipplings are also observed in the soft tissues immediately adjacent to the normal site of the femoral epiphyses (Fig. 3). The sacrum and coccyx share abundantly in this punctate appearance (Fig. 4).

6. The wrists and ankles imitate the paint-brush stippling in the carpal and tarsal components, with a tendency towards the flexed position. Some of these bones show premature ossification (Fig. 5). Cretins may sometimes show mottling and irregularity of the epiphyses, but these occasional findings are quite distinct from the more discrete and refined appearance of the punctate dysplasic epiphyses. The metaphyseal ends are broadened and not bevelled as is observed in Fig. 1.

Literature

A rare disorder of infancy is the presence of discrete centres of dense bone in many of the cartilaginous epiphyses and apophyses.

Hünermann[1] first reported this condition in 1931, since which time 8 cases have been collected by British investigators and, according to Fairbanks,[2,3] altogether 16 such cases have been reported in the literature from all centres. No evidence of any hereditary influence has been described and both sexes appear to be equally affected. The condition begins in foetal life and of the 16 cases collected, 14 were discovered in the first 9 months after birth.

The cause of the condition remains unknown and no late reports are available to enable investigators to prognosticate the course of the condition. Presumably abnormalities in the bone epiphyseal and metaphyseal structures will, if the patient survives, lead to such disabilities as spasm and arthritis. Flexion and contractures occur mainly in the region of the elbow joint, and ulnar deviation results. As mentioned before, cretins may share these deformities but the epiphyseal appearances are different, and the presence of cataracts in the dysplasic infants presents a distinctive feature.[4]

Some cases simulate a Perthe's disease or a dislocation of the hip. Of the 16 cases recorded, over 50% have died, mostly as a result of intercurrent infections of the lungs and the kidneys. Miliary tuberculosis, as superadded pathology, has been responsible for a number of early terminations of life.[5] These observers also found in their autopsies small nodular calcifications in the subcutaneous tissues. Harris[6] has reported mucoid degeneration with cystic spaces in the cartilaginous epiphyses, particularly near the outer epiphyseal surfaces. Some of these were

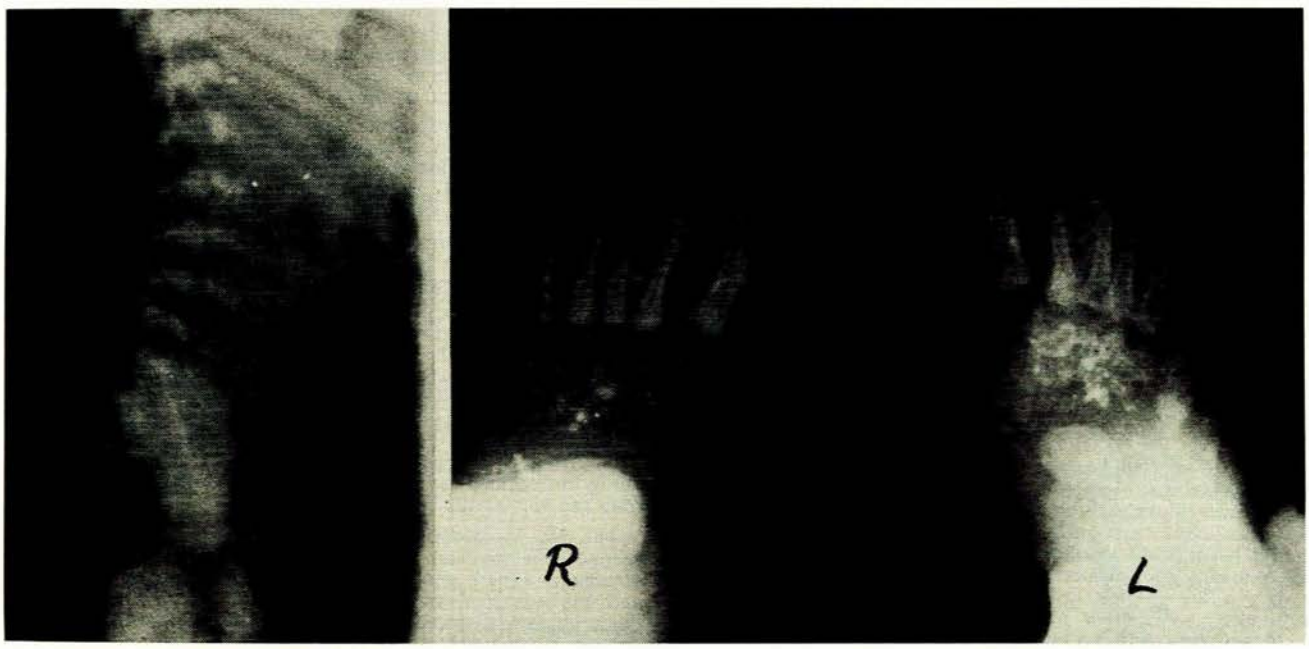

Fig. 4 *Fig. 5*

Fig. 4. Spotted appearance of the vertebrae, sacrum and coccyx. *Fig. 5.* Stippled tarsal bones in right foot and premature ossification in left foot, which is flexed.

invaded by blood-vessels showing similar changes to those found in achondroplasia. This autopsy finding was supported by Lund[7] who found tough fibrous tissues invading the muscles of his patient who died at the age of 4 months.

The infant described in this case report is under the care of the paediatricians at Karl Bremer Hospital. They are watching the progress and are keeping the patient under constant observation.

SUMMARY

1. A case of the rare disorder dysplasia epiphysialis punctata in an infant is described.

2. The condition begins in foetal life, but, as far as has been investigated, the cause of the disorder is unknown. There is no evidence of any hereditary influence.

3. Multiple 'paint-brush stipplings' occur at the epiphyses, and deformity of the hips, elbows and shoulders generally develops.

4. The infant is usually dull generally, has congenital cataracts, and life tends to be terminated at an early age.

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