AN UNUSUAL CASE OF METAPHYSIAL DYSPLASIA

S. D. THEODOROU, ATHENS, GREECE, AND J. ADAMS, CAPE TOWN, SOUTH AFRICA

From the Princess Elizabeth Orthopaedic Hospital, Exeter, England

Cases of metaphysial dysplasia have been recorded by Pyle (1931), Bakwin and Krida (1937) and Hermel, Gershon-Cohen and Jones (1953). Jansen (1934) described a form of metaphysial dysplasia which he termed metaphysial dysostosis. Similar cases were reported by Cameron, Young and Sissons (1954), Lenk (1956), Maroteaux and Lamy (1960), Evans and Caffey (1958), and Gram, Fleming, Frame and Fine (1959). Brennan and Guarino (1961) reported a case of metaphysical growth disturbance with features differing from the types described by Pyle and Jansen. The case reported here bears some resemblance to that of Brennan and Guarino, though it differs widely in certain respects.

CASE REPORT

A girl of two, an only child, was seen because her mother had noticed that she had bow-legs. Teething was normal and the child had sat up at the age of fourteen months and had walked at nineteen months. The only feature of interest in the family history was that a paternal uncle was reported not to have walked until he was aged seven.

On examination the child was under size for her age and her behaviour was like that of a child of twenty months. There was slight bowing of both tibiae and she had a waddling gait. Dental caries was marked.

At the age of three dullness of the hair was noticed and thyroid extract (1/4 grain daily) was administered for one year and an increased dosage (1/2 grain daily) for a further year.

The child remained under observation for eleven years, and was admitted when aged fourteen because of low back pain and vague pains in the legs. She showed evidence of mental and physical retardation. The mental age was about nine years. She was short for her age (4 feet 9 inches) and she weighed 79 pounds. She stood with increased lumbar lordosis (Fig. 1) and the gait was slightly waddling. Mobility of the spine was full and the upper limbs appeared normal. The hips showed 5 degrees of flexion deformity on each side and flexion was limited by 10 degrees and abduction by 20 degrees. There was some varus deformity of the lower femora and the legs below the knee appeared thickened (Fig. 2). The genitalia and secondary sexual characters were underdeveloped.

Investigations—Haemoglobin content of the blood was 10.5 grammes per cent. The red cells showed slight anisocytosis. Reticulocytes comprised 1 per cent, and there were 202,000 platelets per cubic millimetre. White cells totalled 3,800 per cubic millimetre.

FIG. 1
The patient aged fourteen, showing increased lumbar lordosis (Fig. 1), bowing of the lower ends of femora and thickening of the upper ends of the tibiae (Fig. 2).

FIG. 2
The patient aged fourteen, showing increased lumbar lordosis (Fig. 1), bowing of the lower ends of femora and thickening of the upper ends of the tibiae (Fig. 2).
FIG. 3
Radiograph of the hips at the age of four, showing triangular fragments in the inferior aspect of the femoral necks.

FIG. 4
The hips at the age of eleven. The triangular fragments have been incorporated but the structural change in the metaphyses is still marked with thickening of the trabeculae, irregular calcification and varus deformity of the femoral necks.
millimetre (neutrophils 26 per cent; lymphocytes 66 per cent; monocytes 8 per cent). Serum calcium was 10·9 milligrams per cent, serum phosphate 5·1 milligrams per cent, and alkaline phosphatase 33·2 units per cent. Electrolyte estimations showed sodium 135 milli-equivalents per litre, potassium 4·3 milli-equivalents per litre, and chloride 107 milli-equivalents per litre. The carbon dioxide combining power was 21·4 milli-equivalents per litre.

Sternal marrow puncture showed normal cells but there was slight depression of haemopoiesis and lymphocytosis. The blood urea was 17 milligrams per cent. The total urine volume was 450 millilitres in twenty-four hours (total calcium 0·126 grammes, total inorganic phosphorus 0·684 grammes). The urea clearance test was 100 per cent of normal. The amino-acid chromatogram showed a normal picture.

Intravenous pyelography showed satisfactory excretion with normal filling of the calyces at five minutes. **Radiographic examination**—The radiological changes were confined to the lower limbs and consisted of lesions involving the femoral neck and the metaphyseal regions of the lower ends of the femora and upper ends of the tibiae. The changes were seen mainly on the inner aspects of the bones, the epiphyses being unaffected. The progression and final outcome of these changes is well shown in the series of radiographs taken over eleven years. In the early stages there was a disturbance of the metaphysis with the apparent separation of a large triangular fragment on the inferior aspect of each femoral neck (Fig. 3). These appearances altered with time, the triangular fragment uniting with the main part of the neck on both

![Radiograph of the knees at the age of eight, showing the metaphyseal defects confined to the inner sides of the lower femoral and upper tibial metaphyses, with disturbance of the normal trabecular pattern, irregularity of the adjacent cortex and bowing of the femoral shafts at the lower end. The epiphyses are all normal in appearance.](image-url)
sides, and the femoral necks becoming thickened and shortened until the final healed stage was reached, leaving residual coxa vara and loss of normal contour of the femoral necks (Fig. 4).

The lesions in the knees were confined to the medial aspects of the femoral and tibial metaphyses and consisted of loss of normal internal structure, distortion of the outline of the bones and bowing of the lower femoral shafts (Fig. 5). These appearances changed until relatively normal trabeculation was re-established, but the metaphysial regions remained abnormal in contour and showed loss of modelling on the medial aspect (Fig. 6). Radiographs of the skull, spine, and upper limbs were normal, except that the hands showed shortening of the middle phalanx of the little finger on both sides. There was no generalised rarefaction and the cortices of the bones appeared normal.

**DISCUSSION**

Metaphysial dysplasia was first described by Pyle in a boy of four as a skeletal disorder affecting the metaphysial region of tubular bones. The main feature was loss of remodelling, giving rise to characteristic radiographic appearances (Fig. 7).

Bakwin and Krida (1937) reported on the same boy and on his sister who presented with the same condition, under the term familial metaphysial dysplasia. In 1934 Jansen described a boy of ten in whom there was gross disorganisation of the metaphysial regions of the tubular bones associated with deformities and dwarfism. He called this condition metaphysial dysostosis. Lenk in 1956 reported a new case, that of a girl of two with a family history, and called it hereditary metaphysial dysostosis.

---

**FIG. 6**

The knees at the age of fourteen, showing irregularities in ossification on the medial side of the lower femoral metaphyses.
Brennan and Guarino (1961) described a patient in whom there were changes in the lower metaphyses of the femora and the upper and lower metaphyses of the tibiae associated with bow-leg deformity. In all these cases the common feature was abnormality of the metaphysis with a normal epiphysis.

Muller and Sissons (1951) reported a patient who presented with clinical and radiological features of metaphyseal dysostosis, but it was proved histologically to be a case of renal osteodystrophy. The authors questioned whether Jansen's original case was not also one of renal rickets. Anderson, Hodgkinson and Pyrah (1961) described cases of renal osteodystrophy due to congenital abnormality of the bladder neck and urethra and to chronic pyelonephritis in association with renal hypogenensis. The authors stressed the difficulties of diagnosis in early cases.

The case reported here does not resemble any of the cases of metaphyseal dysplasia previously reported but has some features in common with Brennan and Guarino's case.

In Pyle's type of metaphyseal dysplasia there was no radiological evidence of involvement of the internal structure of the metaphysis, the main feature being loss of remodelling of the long bones of the limbs. In Jansen's there was gross derangement of the metaphyses which were expanded and had extensive changes of the internal structure, and marked shortening and deformity of the bones had resulted. In Brennan and Guarino's case the changes were confined to the lower limbs and there was bow-leg deformity. The triangular fragment shown in the upper and lower tibial metaphyses closely resembled the triangular fragment in the medial side of the femoral neck in our case but the changes in the metaphyses were more severe.

Osteochondrosis deformans tibiae (Erlacher-Blount's disease) should be easily differentiated because the abnormality is usually confined to the upper medial epiphyseal region of the tibia. The medial part of the metaphysis and also the epiphysis fail to develop and varus deformity results, with the greatest curvature of the limb at the level of the knee.
AN UNUSUAL CASE OF METAPHYSIAL DYSPLASIA

joint. The rest of the metaphysis and the diaphysis are normal. Occasionally similar changes may be present in the upper and lower epiphyses of the femur, but the picture is quite characteristic as the epiphyses are also affected (Michail, Theodorou and Chouliaras 1959).

We feel that renal osteodystrophy can be excluded. Although the radiological changes in renal osteodystrophy are variable the appearances in our case are quite unlike them. No abnormalities were found in the kidneys and the calcium and phosphorus metabolism was normal. The only feature which we are unable to explain is the pancytopenia.

It would appear that our patient represents a forme fruste of Jansen’s type of metaphysial dysplasia. The metaphysical disturbances were of mild degree with the result that severe deformities of the long bones did not occur. Healing, with the closure of the epiphysial line, was the final result.

SUMMARY

1. A case of metaphysial dysplasia observed for eleven years is reported.
2. Only one case with similar features has previously been reported.
3. The literature on the subject is reviewed.

The patient has been under the continuous observation of Mr Norman Capener. We present the case here with his permission, and owe him our thanks for helpful criticism and advice.

REFERENCES


