6. METAPHYSEAL DYSOSTOSIS

In this excessively rare condition the metaphyses of all long bones consist for the most part of unossified cartilage. The radiographic appearances are unique and they differ considerably from those seen in dyschondroplasia.

In 1934 Murk Jansen of Leiden published a case. Seven years before he had presented a set of radiographs of the patient, who was then five years of age, to the Royal College of Surgeons of England. By courtesy of Sir Arthur Keith, formerly Conservator of the Hunterian Museum, the author was able to secure prints of all the films. Search of the literature has failed to disclose a single comparable case.

The patient was a boy, born with club feet which were treated by manipulation. The ankles, and the lower ends of the radii and ulnae, gradually thickened. The child crawled at six months, but he was unable to walk at three years. Eventually, however, he succeeded in walking with the help of splints. He was dwarfed, and at ten years of age was at least twelve inches below normal height. The lower limbs were more affected than the upper limbs, and they were markedly deformed. The feet were valgoid. The skull and spine were normal. Dentition was somewhat delayed. At five years there was some anaemia, and the serum calcium was high (16.6 mgm.) but this gradually came down to 13 mgm. per cent. Other investigations of the blood and urine were negative. At ten years further investigations revealed no abnormality. Jansen considered that the cause was abnormal intra-uterine pressure acting at a particular stage of foetal development, a theory which he postulated in the explanation of other developmental errors.

Radiographic features — The epiphyses have developed remarkably well, and are of normal density, but the metaphyses both at five years and ten years of age are "still cartilaginous and irregular, cloudy and impregnated with salts." Some of the epiphyses are displaced from the lines of the shafts. This is more obvious in films taken at the age of five years than later. The shafts of the long bones are stout; many are curved, and they end very irregularly with some expansion. There is a wide interval between shaft and epiphysis, the space containing a varying number of rather dense and discrete centres of ossification, especially close to the shaft. There is no suggestion of a columnar arrangement of the cartilaginous metaphyses as in dyschondroplasia, and the epiphyses are not mottled, either partly or wholly, as they may be in the allied condition. The spots of calcification and ossification in the metaphyses vary in size, shape, and density, and they are not quite like the circular dense spots which are seen in the "healing stage" of dyschondroplasia. Many of the metaphyses appear to be enlarged. Changes in the hands differ strikingly from those of dyschondroplasia; the cartilage masses, dotted with fragments of bone, are seen only adjacent to every epiphysis and they are not distributed irregularly as isolated enchondromata anywhere in the shafts of the metacarpals and phalanges. The carpus and much of the tarsus seem to be ossified normally, but the posterior part of the os calcis shows changes similar to those in a metaphysis. At the age of five years, but not at ten years, irregular or punctate ossification is seen in the navicular and cuneiforms. In the pelvis the anterior parts of the ilia are mottled, but there is no such motting, as one might have expected, in the bone near the crests. The skull, spine, ribs, and clavicles appear to be ossified normally.

Pathology — A fragment from the lower end of one tibia, including portions of the shaft and the cartilaginous metaphysis, was examined histologically. The cartilage cells were either small and collected into nests, or large and "myxœdematous." Calcification was seen in the matrix. The junction of diaphysis and cartilage was irregular, and isolated masses of bone were seen in the substance of the cartilage, "as in rachitis."

REFERENCE

Case 39. The boy at the age of ten years, showing deformities of the lower limbs. Fig. 113 is a radiograph of the right shoulder joint and upper arm at the age of five years, showing the unossified cartilaginous metaphyses at both ends of the stout humeral shaft, the mottled centres of ossification at the upper end, and the well-ossified epiphyses at both ends. Fig. 112 shows the dwarfing, he was twelve inches below normal height.
Case 39. Hands at the age of ten years, showing the changes not scattered throughout the shafts as in dyschondroplasia, but concentrated almost entirely in the metaphyses. Note that the carpal bones are unaffected.

Case 39. At the age of five years, showing irregular ossification of the anterior part of the ilia and the acetabula, and the stout curved femora with expanded upper extremities and largely unossified necks.
Case 39. Ankles and feet at the age of ten years showing very irregular ossification and deformity of the lower metaphyses of tibiae and fibulae. Note well-formed epiphyses of these bones and irregular ossification of posterior part of the os calcis in both feet.

Fig. 116

Case 39. Knees at the age of five years, showing the stout femora, the unossified metaphyses with mottled centres near the diaphyses, and the well-ossified epiphyses of both the femora and tibiae. Note the displacement of the upper tibial epiphyses.

(Figs. 112, 114, and 116 are taken from Dr Jansen’s article.)