DIAPHYSIAL ACLASIS

Report of an Unusual Case

V. H. ELLIS and J. G. TAYLOR, LONDON, ENGLAND

From the Orthopaedic Department, St Mary's Hospital

This case of diaphysial aclasis is described because it presents three unusual features: 1) sudden spontaneous growth of one of the exostoses, clinically malignant but not confirmed as such histologically; 2) associated congenital abnormality of the cervical vertebrae; 3) radiological appearance of some of the bones which might cause confusion with dyschondroplasia.

CASE REPORT

The patient was a factory gatekeeper aged forty years, whose mother had first noticed numerous hard swellings in his limbs and on his chest when he was three years old. At the age of fifteen, during the course of a year, his left arm became almost completely paralysed below the elbow, with loss of sensation; and at about this time two of the bony swellings in the left arm were removed. Eighteen months before admission, a swelling which had always been present in the upper part of the left thigh began to grow and a few months later it became painful. He came into hospital because of difficulty in getting his trousers over the swelling. None of the other bony swellings had shown any increase in size since he stopped growing, and his general health had been good.

Family history—A similar condition was present in three successive generations (Fig. 1).

Examination—His general condition was satisfactory. His height, sixty-two inches, was below average. Bony tumours and deformities were widely distributed as the photograph (Fig. 2) and radiographs (Figs. 3 to 7) show. The left side of the body was more severely affected than the right. The tumour of the left thigh was twenty-nine inches in circumference and eleven and a half inches in its vertical length. It was hard; the skin was stretched tensely over it but was not adherent; the skin temperature was not increased. Mechanical interference by the tumour limited flexion at the left hip joint by 40 degrees. The chest was deformed with two antero-lateral sulci, the right deeper than the left. Most of the sternal head of the left pectoralis major was absent (Fig. 2). There was a thoracic kypho-scoliosis of the spine with convexity to the right, with slight restriction of movement in all directions and

\[ \delta = \bullet \text{Died, aet. 80 years} \]

[Diagram of family tree]

Aet. 63 years, alive and well

This patient

Aet. 13 years.
Radiographs normal

\[ * \text{Thought to be normal but examination refused.} \]

Fig. 1

Family history. The black signs indicate the members of the family affected by diaphysial aclasis.
FIG. 2

Photograph of the patient showing the multiple swellings, gross enlargement of the left thigh, absence of most of the sternal part of the left pectoralis major muscle, and the thoracic deformity.

FIG. 3

Figure 3—The left femur. Dense areas of calcification are scattered throughout the large tumour mass.

FIG. 4

Figure 4—The left knee joint, showing marked overgrowth of the medial tibial condyle capped by a large densely calcified chondroma.
Fig. 5
Calcified chondroma arising from the third left lumbar transverse process.

Fig. 6
Figure 6—Cervical spine, showing fusion of the bodies and of the spinous processes of the fourth and fifth, and the sixth and seventh, vertebrae.

Fig. 7
Figure 7—The hands and wrists. In the right hand there is lack of modelling of the phalanges and metacarpals. The lower end of the left ulna is ill-formed and stunted, a well recognised feature of diaphysial aclasis.
considerable limitation of cervical rotation. The left clavicle was bowed over a tumour, two inches in diameter, which arose from the left first rib and had caused a left brachial plexus lesion of lower trunk type (C.8 and T.1) affecting the left arm. The cardio-vascular and respiratory systems were normal, except for limited thoracic respiratory movement. The liver edge was palpable two fingers' breadth below the costal margin but no irregularity could be felt on its surface. The blood picture was normal.

It was considered that malignant change had occurred in the previously benign tumour in the left thigh. As there was no evidence of metastases, disarticulation at the hip was undertaken. It was just possible to ligate the vessels immediately below the inguinal (Poupart's) ligament and to perform the disarticulation without cutting into tumour substance.

After operation the patient's condition was satisfactory until the second day when, after a standard dose of morphine derivative, he became comatose for several hours. Next day, his condition again appeared satisfactory, but on the fourth day he complained of abdominal pain, his condition steadily deteriorated and he died.

_Autopsy_—The cause of death was peritonitis from perforation into the lesser sac of a small acute peptic ulcer on the greater curvature of the stomach near the cardia. Thorough examination of the whole body revealed no secondary neoplastic deposits. The tumour of the left thigh was cartilaginous in nature. It involved the shaft of the femur and was adherent to the surrounding muscles which were partly fibrosed (Fig. 8).

_Histological examination_ (Professor W. D. Newcomb)—There was no unequivocal evidence of invasion of the muscle but several small outlying nodules of tumour appeared to invade
the fibrous tissue. The cytology of the tumour varied from large degenerate cartilage cells to small spindle and angular cells forming reticulin and collagen. Some cells were binucleated but no mitotic figures were seen. It was impossible on histological grounds either to confirm the clinical suspicion of malignant change or to prove that the tumour was benign.

**DISCUSSION**

In this case the rapid increase in size of a pre-existing exostosis, with pain, many years after general cessation of bone growth, strongly suggested malignant change, and radical surgery was clearly indicated. The diagnosis of malignancy in diaphysial aclasis must be made chiefly on clinical grounds because of the uncertainty of histological diagnosis of chondrosarcoma. Increase of symptoms in an adult—renewed growth and pain—must generally be regarded as evidence of malignancy. Platt (1931), however, stated that rapid growth and large growth are not in themselves pathognomonic of malignant change in a chondroma. Jaffe (1943) considered that malignant transformation in diaphysial aclasis took the form of chondrosarcoma, not of osteogenic sarcoma: chondrosarcoma may originate in the cartilaginous cap of an exostosis or its residuum, or from a subperiosteal cell-rest. Jaffe and Platt both mentioned the difficulty of histological diagnosis. Chondrosarcoma arising in diaphysial aclasis is not usually highly malignant; it tends to remain only locally invasive for some years; distant metastases occur late and in many cases not at all (Jaffe 1943, Geschickter and Copeland 1949, Willis 1948, Gardner 1937).

The incidence of malignant transformation is difficult to determine because so many minor cases of diaphysial aclasis are not seen by surgeons. Jaffe found 11 per cent (three out of twenty-eight cases). Bennett and Berkheimer (1941), Gardner (1937), on the other hand, thought that it was rare. It almost certainly occurs more often in adults than in children.

The recommended treatment of clinically diagnosed malignant change in an exostosis is local resection, if technically possible; amputation at the site of election above the tumour is necessary in advanced growths. X-ray therapy is of doubtful value. It should be tried in inoperable growths or if operation is refused.

*The abnormality of the cervical vertebrae*—Fusion of vertebrae, as in this case (Fig. 6), is not one of the recognised features of diaphysial aclasis. Fairbank (1949) stated that exostoses may affect the neural arches but seldom the vertebral bodies; he did not mention fusion. Keith (1920) stated that in diaphysial aclasis all bones formed entirely within cartilage or entirely within membrane—the tarsal and carpal bones, the epiphyses of the long bones, the sternum, the vertebral bodies, and the bones of the cranial vault and face—are free from the disorder; it is confined to those parts of the skeleton where bone laid down in cartilage becomes covered with periosteal bone. He considered that there was an arrest of development in the periosteal ring (or ferrule) which permits the cartilage of the growth disc to be exposed on the surface, thus leaving it uncovered and free to give rise to the exostoses. The question arises then whether the fusion of the cervical vertebrae in this case could be part of the generalised diaphysial aclasis, or whether it was a separate and coincidental congenital abnormality. According to Stocks and Barrington (1925) bones formed strictly in cartilage or in membrane are not invariably spared, as stated by Keith, but are occasionally the site of an exostosis. If so, fusion of vertebral bodies might be part of the disorder. These authors also mention the occasional occurrence of scoliosis or kyphosis, or both, and thoracic deformities similar to those of rickets, which abnormalities were present in this case.

*Differentiation of diaphysial aclasis from dyschondroplasia*—The radiographs in this case showed more calcified cartilage than that usually seen in diaphysial aclasis. This might suggest dyschondroplasia, in which speckled calcification occurs in the affected areas of the diaphyses and epiphyses as the subject grows older (Hunter and Wiles 1935). But the masses of calcified cartilage sometimes seen in diaphysial aclasis are not endosteal as in dyschondroplasia;
they are caused by local disturbances of endochondral ossification in the proliferating cap of hyaline cartilage overlying the exostosis (Jaffe 1943) (Fig. 9). Small exostoses may arise near the epiphyses of the long bones in dyschondroplasia, but they are rough and irregular, may point in any direction and do not grow large; they are unlike those of diaphysial aclasis. In dyschondroplasia the expansion of the ends of the long bones—a significant feature of diaphysial aclasis—is very seldom seen.

The asymmetrical distribution of the lesions in our case might also suggest dyschondroplasia. In fact, however, dyschondroplasia may be symmetrical or asymmetrical; and in diaphysial aclasis, though the distribution of exostoses is usually fairly symmetrical, it is not always so. There is no evidence of a hereditary factor in dyschondroplasia. Radiographs of the hand showed only defective modelling of the phalanges and metacarpals. There were no enchondromata such as may occur in dyschondroplasia. Jaffe (1943) maintained that dyschondroplasia and diaphysial aclasis are unrelated, because the two conditions do not occur in members of the same families, and enchondromata do not occur in association with exostoses; he suggests that faulty interpretation of radiographs (perhaps taken only in one plane) may be the cause of confusion between the two conditions.

REFERENCES

Jaffe, H. L. (1943): Archives of Pathology, 36, 335.