DIAPHYSEAL ACLASIS: REVIEW OF LITERATURE AND REPORT OF AN UNUSUAL CASE

(A MASSIVE VERTEBRAL OSTEOCHONDROMA WITH COMPLICATIONS)

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This condition was first described as a clinical entity by Virchow[1] under the name of 'exostosis multiplex cartilaginea'. Keith[2] contributed largely to its recognition and description in England and was the first to name it 'diaphyseal aclasis'. The word 'aclasis' was suggested to Keith as a descriptive title of the disease marking the 'failure' of the periosteum to model the bone properly. In America Ehrenfried's name, 'hereditary deforming chondrodysplasia' is sometimes adopted.[3] Another common name is 'hereditary multiple osteocartilaginous exostosis'.

Ollier's disease[4] (multiple enchondromata; dyschondroplasia) is now regarded as a completely separate entity by most authorities.[5-7] Some authors[8-10] have reported cases which have appeared to present features of both diaphyseal aclasis and Ollier's disease, but these accounts of the supposed association of these conditions are not at all convincing. Most people will agree with Fairbank's comment[6] that the 2 conditions are frequently confused, but with little if any justification — they are radiologically and in other ways quite different, and in the vast majority of cases are readily distinguishable.

This paper is the report of a case of multiple exostoses with most marked complications.

Diaphyseal aclasis is characterized by the presence of multiple cartilage-covered exostoses and calcified cartilaginous outgrowths throughout the skeleton; in Ollier's disease islands of cartilage remain unossified within the shaft, giving rise to enchondromata.

PATHOLOGY

The aetiology of diaphyseal aclasis is not known and many theories have been advanced. This is not surprising, since the question of the cause of this condition involves the fundamental problems of heredity and of bony growths.[11]

Keith[2] considers there are 2 abnormal processes: (1) One or more fragments of cartilage from the epiphyseal line, becoming isolated on the surface of the metaphysis, proliferate and form tumours; (2) the periosteum, which is incomplete at the sites of these cartilaginous nests, fails to model the metaphysis in a normal way.

The lesions are often not discovered until childhood or adolescence, but they have been identified radiologically at birth.[6]

Distribution

The exostoses project from the surface of the metaphyses, or adjacent diaphysis,[22] and are common near the rapidly growing ends of these bones.

The long bones, the pelvis and scapula are chiefly affected. Many of the patients suffering from this condition are below normal height owing to shortening and bowing of the long bones. Unlike Ollier's disease, the hands are normal[18] or the metacarpals and phalanges may show small spikes projecting from the cortex. Common sites for these lesions are the region of the knee joint, lower ends of the radius and ulna, and the upper ends of the femur and humerus respectively. The elbow-joint region is usually free from changes. The pelvis and scapula are frequently involved, often giving rise to large lobulated cauliflower-like tumours which show no differentiation into cortex and medulla. These lesions are then regarded as osteochondromata. The ribs, clavicle, sternum, mandible and skull show outgrowths less frequently.[19] The carpal and tarsal bones and the spine are only rarely involved.[20] In the spine it is usually the neural arches that are affected and occasionally the vertebral body. These lesions are said to be small; Fairbank[6] has seen only 2 cases with large exostoses of the spine.

Complications

Because of their size most of the exostoses cause no symptoms at all. The commonest complications of this condition are:

1. *Deformity.* This results especially from shortening of the ulna, either with or without bowing of the radius, interference with joint mobility and with muscular and tendon activity.

2. *Pressure symptoms.* A case with involvement of cranial nerves, including the optic nerve, was reported by Nielson.[12] Multiple pelvic exostoses may complicate the delivery of a child during labour.[13]

3. *Malignant degeneration.* A certain number of cases develop malignant changes in one of the lumps. Geschickter and Copeland[14] reported this in 7% of their cases; Jaffe[5] has estimated the incidence of sarcoma as over 10%: it occurred in 3 of a series of 28 cases, and he pointed out that the incidence of malignancy might ultimately be much

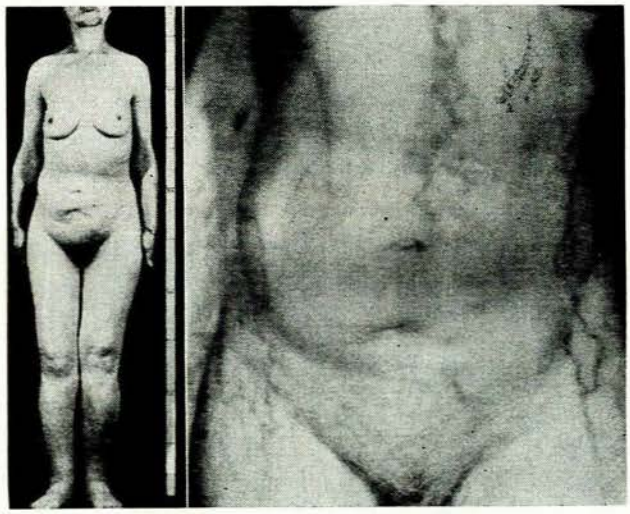

Fig. 1. Showing the patient with a vertebral osteochondroma presenting on the right side of the lower abdomen. Note collateral veins of the abdomen and lower limbs.
Fig. 2. An infra-red photograph demonstrating multiple collateral veins of the anterior abdominal wall, flanks and lower extremities as a result of obstruction of the inferior vena cava.

higher than this, since the majority of his cases were still young when the survey was made. Malignant change may be suspected clinically if a lesion becomes painful, or increases in size. Normally the growth of an exostosis stops

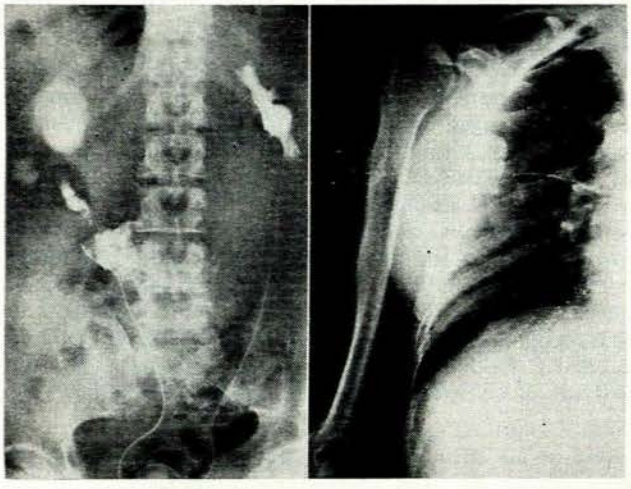

Fig. 3. Retrograde pyelogram showing deviation of the left ureter, rotation of the left kidney, displacement and obstruction of the right ureter with hydro-ureter and hydronephrosis. Note the large vertebral osteochondroma showing typical punctate calcification and probable sarcomatous change.
Fig. 4. Lesions seen in the upper shaft area of the right humerus, fourth right rib, and upper border of the right scapula — showing irregular calcification.

when skeletal growth ceases — continuation of growth of such a lesion after the age of 20 - 22 is suggestive of malignancy. A chondrosarcoma may start in the cartilage cap of an exostosis. Bones most likely to be involved are the pelvis, femur and scapula.

CASE REPORT

L.V., a female aged 50 years (Fig. 7), was admitted to hospital on 17 July 1958, complaining of a lump in the right groin of 9 months' duration and a constant pain of similar duration in the lumbar region of the spine, right buttock and lateral surface of the right thigh extending down to the knee. She also noticed 'pins and needles' along the lateral side of the right thigh and leg down to the ankle. She could only manage to walk for 5 minutes, when the pain forced her to stop for a while.

A hysterectomy was performed 12 years previously for fibromyomata. She also had some frequency of micturition. No family history of bone disease could be obtained.

Clinical Examination

A short middle-aged patient with an indirect inguinal hernia on the right side, a subumbilical midline scar on the abdomen and multiple collateral veins extending from the groins upwards in the direction of the axillary regions, with the flow directed upwards (see infra-red photograph, Fig. 6).

There was oedema of both lower limbs at the ankles with varicose pigmentation more marked on the left side. Palpation of the abdomen showed a hard, lobulated, fixed and painless mass 10 inches by 6 inches in diameter, extending from the left hypochondrium to the right iliac fossa and into the pelvis. The lower border of the mass could be felt on rectal examination in the hollow of the sacrum. The left kidney was just palpable. No ascites was found. Neurological examination showed hypoaesthesia of the anterolateral surface of the right thigh and medial surface of the right leg, corresponding to the distribution of the second, third and fourth lumbar nerves, in addition to an absent right-knee reflex (L 3 and 4).[15]

There was true shortening of the left lower limb and the right leg (from the antero-superior iliac spine to the medial malleolus) being 33½ inches, compared with 32 inches of the left leg. The distance from the medial malleolus to the knee joint level was: right — 14¼″; left — 13¾″. Height: 5 ft. 3 ins. Weight: 122 lb. BP 210/115 mm.Hg.

Special Investigations

ESR (Westergren) was 15 mm. in the first hour; WR and Kahn tests were negative; CSU = N.A.D.; blood urea = 42 mg./100 ml.; serum alkaline phosphatase 13 KA units; Blood calcium and phosphorus normal.

X-ray findings: A plain X-ray of the abdomen showed a large vertebral osteochondroma (probably sarcomatous) arising from the 4th lumbar vertebra. The main features suggesting malignancy were: (1) continuation of growth giving rise to an increase in size of the lesion, (2) the presence of a large soft-tissue mass and (3) scattered areas of irregular calcification in the soft-tissue mass.

She was operated on a week after admission to hospital for the inguinal hernia, when a laparotomy was performed through a right paramedian incision and a biopsy taken from the large retroperitoneal mass. Histologically the specimen for biopsy consisted of hyaline cartilage with cells of varying size; the report stated that no evidence of malignant degeneration was present in the portion removed for section. A survey of the skeleton demonstrated multiple other lesions. Lesions were found on all the bones of the skeleton except the skull, carpal and tarsal bones.

IVD showed rotation of the left kidney, a non-functioning kidney on the right side, and indentation and deformity of the bladder, caused by the very large vertebral lesion.

A retrograde pyelogram (Fig. 3) showed deviation of the left ureter, displacement and obstruction of the right ureter, with a marked degree of hydro-ureter and hydronephrosis.

A chest X-ray showed no evidence of metastases.

The patient has been followed-up for 18 months post-operatively with no evident change in her condition.

DIAGNOSIS

Radiography

Diagnosis is usually made radiologically. In every case

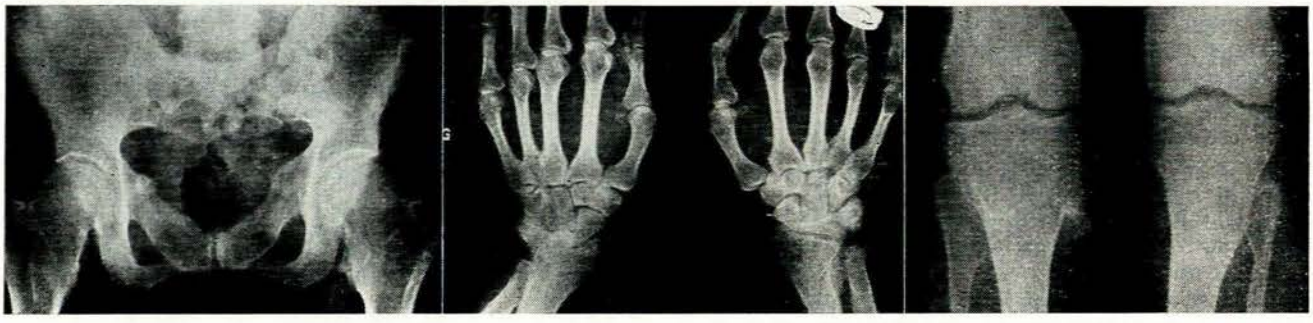

Fig. 5. Pelvis and hip joints showing exostoses in the region of the femoral necks, ischial spines and left sacro-iliac joint.
Fig. 6. Wrists showing cartilaginous exostoses with bilateral shortening of the ulnae as well as irregularity and obliquity of the articular surfaces. Note slight deformity of the shaft of the 5th lumbar metacarpal and spikes of phalanges.
Fig. 7. Knee joint showing exostoses of femora, tibiae and fibulae.

of disease of the end of long bones or of bones of the hand or foot, radiograms of the entire skeleton should be taken, otherwise a case of diaphyseal aclasis or Ollier's disease may be overlooked.[23]

There are 3 salient features:

1. *Formation of multiple exostoses of the pedicle or broad-base type*. The exostoses vary from simple spikes to pedunculated or large cauliflower osteochondromata (Figs. 3 and 4).

2. *Widening of the metaphyseal side of the long bones* (as seen at the upper end of the femurs especially on the left side in Fig. 5). This is usually best seen when there are numerous exostoses present.

3. *Deficient growth of the ulna* (seen bilaterally in Fig. 6), *or fibula* (Fig. 7) with continued growth of the radius and tibia producing bowing of the radius or tibia.

The last 2 features are often seen in Ollier's disease.

The phalanges, metacarpals and metatarsals usually show nothing or only small 'spike' exostoses. The minimal changes in the hands in this case is demonstrated in Fig. 6. Pale mottled areas with dense punctate spots are seen in the lumbar osteochondroma.

The long bones — humerus (upper end — Fig. 4), radius, ulna (Fig. 6), femur, tibia and fibula (Fig. 7) usually show the typical appearances.

Brailsford[16] remarked on the common finding of irregularity and obliquity of articular surfaces (Fig. 7).

DIFFERENTIAL DIAGNOSIS

The diagnosis depends largely upon X-rays. A biopsy should be done in doubtful cases and to help assessing malignant degeneration. However, as in the present case, radiological evidence of malignancy is of much greater importance than a negative pathological finding.

Ollier's disease (*multiple enchondromatosis*) — mixed types may occur. Heredity is less commonly seen. It may be largely unilateral in distribution. There is often gross deformity of the hands. A biopsy may be needed.

Maffucci's syndrome[17] is a condition in which dyschondroplasia is associated with cavernous haemangiomata and phleboliths in the soft tissue.

Albers-Schönberg disease (*marble bones*) usually shows typical X-ray changes of dense bone.

PROGNOSIS

The majority of cases reported in the literature are children. When the patients reach adult life their symptoms will be due to complications. Chondrosarcomata are relatively slow-growing tumours and metastasize late.

TREATMENT

Regular follow-up (both clinical and radiological) is needed, in case malignancy develops. Köhler performed an osteotomy already in 1905 for advanced bowing of long bones.

Surgical treatment of 1 or more of the exostoses may be indicated for cosmetic reasons or on account of interference with joint mobility or pressure on tubed organs. It is important to realize that any osteocartilaginous exostosis which becomes painful or increases in size should be regarded as a chondrosarcoma and treated by surgical removal. This should only be attempted if it is possible to remove the growth with a good margin of normal tissue around it.

In the case presented, it is obvious that surgery of the vertebral osteochondroma would be a hazardous undertaking.

Radiotherapy — supervoltage radiation in inoperable chondrosarcoma may give some palliation.

SUMMARY

A case of multiple exostoses (diaphyseal aclasis) has been presented in a middle-aged woman. There are two vertebral cartilaginous exostoses arising from the 5th cervical and the 4th lumbar vertebrae. The latter was very large (10 in. x 6 in.); it had apparently undergone malignant degeneration, and caused obstruction of tubed organs (ureter and inferior vena cava) and compression of nerves (L 2, L 3 and L 4). Technical difficulty and size of tumour made it inoperable. Fairbank,[25] up to 1951, saw only 2 patients with large exostosis of the spine.

The case also presents bilateral shortening of the ulna with minimal changes in the hands.

Review of the literature shows the need to distinguish carefully between multiple exostoses (diaphyseal aclasis) and multiple enchondromatosis (Ollier's disease).

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