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SARCOMATOUS CHANGE IN THREE BROTHERS WITH DIAPHYSIAL ACLASIS

BY

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Sarcomatous change is a rare but well-recognized complication of diaphysial aclasis which has never been recorded in two members of the same family. In the family described here, three affected brothers have died of bone sarcomas in the past 25 years.

"Diaphysial aclasis" (Keith, 1920) is the name commonly used on this side of the Atlantic, but there are several synonyms. Ehrenfried (1915) used the term "multiple cartilaginous exostoses," which has become recognized in the United States of America. Jaffe (1943) and Monro and Golding (1951) called the condition "hereditary multiple exostoses." "Multiple exostoses" was used by Abernethy (1830), Virchow (1876), and MacCallum (1940).

Two abnormalities of bone growth characterize diaphysial aclasis. There are osteochondromas, which are more numerous near the epiphyses of the long bones, which grow most actively (Fairbank, 1949, 1951). However, they have been described on all bones, including those of the face and skull, which are preformed in membrane (Honeij, 1920). The exostoses lie between the bone and the periosteum, and show no tendency to erode the parent bone. The ossification of the epiphysial plates of affected bones is disordered. Instead of the normal "row formation" of the epiphysial cells (Dodds, 1930; Johnston and Whillis, 1938), there are masses of cartilage of unusual thickness and patches of ossification. The general appearance is of uncontrolled cartilaginous growth, but without proper preparation for the deposition of cancellous bone. The affected bones fail to grow to their proper length, and the metaphyses are expanded into an irregular cylindrical shape.

The exostoses vary in number from one to 1,000. They appear at any time in childhood or adolescence, and continue to grow until bone-growth ceases at about the age of 20 years. After this they remain static unless malignant change occurs. By the age of 10 years 90% of exostoses have been discovered (Stocks and Barrington, 1925). The unequal shortening of the long bones of the legs causes an affected person to be below average height and to have abnormalities of stance such as kyphoscoliosis, coxa vara, knock-knees, and flat-feet. In middle age severe osteoarthritic changes are frequent in the weight-bearing joints. Bessel Hagen (1891)

described the shortening of the distal end of the ulna with a tendency to dislocation of the wrist, which is said to occur in a third of affected people.

Family History

Eight members of this family have lived in Hoddesdon (Numbers 2, 3, 4, 7, 14, 15, 16, 17), and most of their relatives live in the Mons areas of Belgium. I was able to interview 21 blood relatives, and all except two of them allowed me to make a full clinical examination. The oldest living relative (Case 1) denied all knowledge of the family complaint, but she allowed me to palpate her arms and legs. I found exostoses around her knee-joints and osteoarthritic changes in these joints. Five of her seven children had diaphysial aclasis, and three of them have died from bone sarcomas. From the family tree (see next page) it appears that the common female ancestor, who is marked "R," must have carried the gene for diaphysial aclasis. Thirty-two of her direct descendants have been traced; 19 were examined fully; two were interviewed; seven were not seen personally; and four were dead at the time of this survey.

In this family there were seven affected males and six affected females. Ten females and nine males were not affected, but it is likely that some of the younger children will develop exostoses as they grow up. Adults with a very small number of exostoses may have been overlooked, as it was not possible to arrange for x-ray films to be taken of all members of the family.

All the affected persons had an affected parent. There was no instance of diaphysial aclasis being inherited through an unaffected parent of either sex. The 13 affected people have had a total of six affected male and four affected female children, as well as five unaffected children of each sex. Exactly half the children of the affected parents have diaphysial aclasis.

It was the "lumps on the bones" which the patients had noticed, and about which they talked. On the whole they had caused very little inconvenience. Only in Case 8 had an exostosis been removed from the lower end of his right radius, at the age of 11 years. The patient's right wrist had been weak since the operation as there was shortening of his radius, which may have been due to damage to the growing epiphysis. In no case was there any symptom from pressure of a benign exostosis on to a joint, tendon, artery, vein, nerve, pelvic outlet, or any other structure. In the nine cases examined the number of exostoses varied from 8 to 78, and were found on most of the bone that had been preformed in cartilage. No exostosis was found on a "membrane-bone." Exostoses were found around the knee-joints of all affected persons. Where an exostosis could be palpated it was usual for x-ray examination to reveal several other small nodules. In Case 7, who was 14 years old, the exostoses on her scapulae appeared larger by palpation than on the x-ray film. In this case the bulk of the exostoses must have been composed of radiolucent cartilage.

None of the patients had noticed the abnormal bone formation at the metaphyses. The cylindrical section and the abnormal internal structure of the affected metaphyses were easily seen on the x-ray films. None of the affected people were above average height, and in four cases (Nos. 2, 7, 10, and 12) one leg was 1 in. (2.5 cm.) or more shorter than the other. In four (Cases 1, 10, 18, and 20) out of seven of the affected people over the age of 40 osteoarthritis of the knees was very troublesome. Case 20 had well-marked coxa vara, knock-knees, and flat-feet, but these may have been due to healed rickets as she had been born near Mons in 1915, and had spent her childhood in this war-stricken area.

Case Histories

Case 2.—In 1956 this man died of chondrosarcoma of the ilium at the age of 54. He had always been aware of

numerous bony nodules on his body, but they had not altered since he was 20. At 42 and 46 years he had become worried lest the nodules on his femora were growing. X-ray films at these times confirmed the presence of numerous exostoses, but there was no evidence of malignant change. When he was 51 his wife noticed a new lump in his left buttock. It was the size of a hen's egg. There was no pain or tenderness in the lump, but his left leg ached slightly. Biopsy confirmed the clinical diagnosis of chondrosarcoma. Two full courses of radiotherapy were given. There was no dramatic tumour response, but it was

Sarcomatous Change in Diaphysial Aclasis

There is a specimen of a chondrosarcoma arising in an exostosis on the crest of the ilium in the Museum of the Royal College of Surgeons of England. Twenty-five other cases have been recorded in the literature, but in no family has there been more than one case. Single cases have been recorded by Weber (1866), Barlow (1895), Fennel (1938), Bennet and Berkheimer (1941), Ellis and Taylor (1951), Monro and Golding (1951), and Gros *et al.* (1957). Phemister (1930), Gardner (1937), and Geschickter and Copeland (1949) each reported two cases. Jaffe (1943) and Drevon *et al.* (1950) each reported three cases of chondrosarcoma complicating diaphysial aclasis. Stocks and Barrington (1925) found records of six instances of malignant change in 1,172 collected case histories.

Chondrosarcomatous change should be suspected in any exostosis that shows renewed growth at any time after the age of 20. In many cases it is difficult to confirm the diagnosis without biopsy. Several x-ray films taken at intervals of a few months show an expanding radio-lucent tumour with patchy secondary calcification, which lies between the bone and the periosteum, and may erode both these structures. Microscopically it is possible to find areas containing many atypical cartilage cells in which double nuclei are common, though mitotic figures are unusual. There is a reduced proportion of cartilaginous matrix (Thomson and Turner-Warwick, 1955). In many cases, where tumours have been clinically malignant, pathologists have

not found it easy to confirm the diagnosis microscopically (Platt, 1931; Warren, 1931). The diagnosis of chondrosarcoma was not included in the "Register of Bone Sarcomata" of the American College of Surgeons until after 1930.

Thomson and Turner-Warwick (1955) found a 50% 10-year survival in unselected collected cases of chondrosarcoma. It is a slow-growing tumour, which metastasizes to the lungs after many years. In Cases 2 and 26 there was no evidence of metastases. As in Case 2, the tumours are radio-resistant, and the only hope of cure is by amputation, which may have to include half the pelvis or the shoulder girdle.

Summary

A family which shows the typical features of diaphysial aclasis is described. In this family three affected brothers have died of bone sarcomas, but in only one of them was a biopsy available. The literature has been reviewed and reports of 26 other cases of chondrosarcoma complicating diaphysial aclasis were found. In none of these families did more than one member show this complication.

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thought that the lump grew more slowly for a time after this treatment. During the remaining three and a half years of his life he needed two chordotomies for the relief of intractable bone-pain, and a cystostomy because of acute retention of urine from an incoordinated bladder. The tumour of his left ilium grew to be 6 in. (15 cm.) in diameter. Anteriorly it pressed upon the abdominal and pelvic organs and posteriorly it ulcerated through the skin to become a necrotic mass. At the time of death there was no evidence of any secondaries, though a post-mortem examination was not allowed.

Case 25.—In 1936 this man died of a sarcoma of his "shin" at the age of 22. His close relations agreed that he had always had numerous bony tumours, but had enjoyed good health. Three years before his death one of the exostoses on his shin began to grow fairly rapidly. The affected leg was amputated and the family were told that he had a sarcoma. He died soon after this operation. No medical notes are available about this case.

Case 26.—This man was 24 when he died of a sarcoma of his pelvic bone in 1946. His close relatives agreed that he had several bony tumours, but he was accepted for the Belgian Army. He was well until nine months before his death. He was treated at the Institut Jules Bordet in Brussels in 1945, and a summary of his case notes is available. He complained of increasing constipation, which necessitated a colostomy. This was due to a bony tumour of his pelvis, which was diagnosed on radiological evidence as an osteosarcoma. No curative therapy was attempted, but a cordotomy was done to relieve his severe bone-pain. There are no records of necropsy, nor of a biopsy of his tumour. There was no evidence of metastases.

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SIGMOID VOLVULUS IN AN AFRICAN POPULATION

BY

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Volvulus of the sigmoid colon is an uncommon surgical emergency in the United Kingdom, and this point has been emphasized by Bolt (1956), who records only five personal cases and 23 others collected from the West Middlesex Hospital, the Bristol Royal Infirmary, and Weston-super-Mare General Hospital between 1946 and 1955. Vick (1932), reporting nearly 7,000 cases of intestinal obstruction occurring in 21 English hospitals between 1925 and 1932, found only 56 cases of sigmoid volvulus. Similarly, in the United States Hilton and Waugh (1951), from the Mayo Clinic, could find only 16 cases treated there during a period of 39 years. On the other hand, other American authors who have included cases occurring among negroes have significantly higher figures (Griffin *et al.*, 1945; Pool and Dunavant, 1951).

In Eastern and Northern Europe, however, volvulus of either large or small bowel is more common. Perlmann (1925) writes of 35 cases of sigmoid volvulus in a series of 200 cases of intestinal obstruction. Bruusgaard (1947) discusses 91 cases treated in one Scandinavian hospital over a period of 10 years. In India, Andersen (1956) found 24 cases among 168 of intestinal obstruction.

This paper describes the experience of sigmoid volvulus gained at Mulago Hospital, the teaching hospital of the University College of East Africa. This is a 628-bed general hospital which provides an emergency service for the African people of Kampala and the surrounding agricultural district, numbering about one million.

From a study of the literature it appears that no other hospital has the opportunity of dealing with such large numbers of patients presenting with sigmoid volvulus.

Incidence

That the incidence of sigmoid volvulus was high has been known at Mulago for a long time. Burkitt (1952) drew attention to this fact, and pointed out that its occurrence was almost entirely confined to one tribe.

The 104 cases of sigmoid volvulus under review were admitted to hospital during the five-year period 1952–6. Only cases proved either at operation or by post-mortem examination are included in this series. A further 18 cases of intestinal obstruction which are thought clinically to have had a sigmoid volvulus were excluded. These cases resolved either spontaneously or after a diagnostic enema.

Table I shows the numbers of cases of volvulus and intestinal obstruction treated during the five-year period. Table II gives the incidence of sigmoid volvulus among

TABLE I.—Figures From Mulago Over a Five-year Period and From 21 English Hospitals Over a Six-year Period

	Mulago	England
Sigmoid volvulus	104	56
Volvulus of transverse colon	2	0
" " caecum	6	35
" " small bowel	21	85
Total volvulus at all sites	133	176
Intestinal obstruction (excluding ext. herniae)	287	3,625
" " (including " ")	1,230	6,892

TABLE II.—Sigmoid Volvulus

	Mulago	England	Rough Ratio Mulago/England
Percentage of all volvulus	78.2	31.8	2.5:1
" " intestinal obstruction (incl. ext. herniae)	8.45	0.81	10:1
Percentage of intestinal obstruction (excl. ext. herniae)	38.1	1.45	25:1

cases of volvulus and cases of intestinal obstruction. For comparison both Tables include Vick's (1932) findings in England.

Table II shows that sigmoid volvulus forms 78.2% of all cases of volvulus, whereas in England cases in this category account for only 31.8%. Even more striking is the relation of sigmoid volvulus to the total number of cases of intestinal obstruction—ten times as many cases occurring. If external herniae, which also have an abnormally high incidence, are excluded, an even more marked difference is observed, the ratio between Mulago and English figures being roughly 25:1.

Double Volvulus.—Twenty cases of "double volvulus" are included among the figures for sigmoid volvulus. This is a clinical entity which does not seem to occur with sufficient frequency outside Buganda to have gained much attention. The double volvulus recognized at Mulago is a compound volvulus of the small bowel and sigmoid colon.

Sex and Tribe.—From Table III it can be seen that males formed 94.2% of the patients. This high figure

TABLE III.—Incidence of Sigmoid Volvulus in Relation to Sex and Tribe

	Male	Female
Baganda	80 (93%)	6
Other tribes	18 (100%)	0
Totals	98 (94.2%)	6