ENGELMANN'S DISEASE: CASE REPORT AND BRIEF REVIEW

I. M. PATZ, M.B., B.CH. (RAND), *Middelburg, Transvaal*, and H. J. J. VAN HEERDEN, M.B., B.CH. (RAND), D.M.R.D. (LOND.),* *Germiston, Transvaal*

Engelmann's disease is a rare clinical entity of which only 23 authenticated cases have been published. It is characterized by sclerosis of the long bones involving chiefly the middle thirds of the shafts of these bones. We present here what we believe to be the 24th case described, illustrating the typical radiological features of the disease along with some special findings.

Engelmann's disease, or progressive diaphyseal dysplasia, was first described by Cumarati[1] in 1922, 7 years before Engelmann's account[2] appeared in print. Griffiths,[3] in an excellent review of the condition (1956), recognized 21 definite cases. Of these 16 were reported in detail, and Griffiths added the detailed account of a further case; the other 5 cases, although more briefly described were sufficiently documented to make the diagnosis certain. A further case has since been described by Stewart and Cole.[4]

CASE REPORT

In March 1955 a 27-year-old unmarried woman was referred for a dysenteric condition of 18 months' duration. She had frequent loose stools daily, containing much blood and mucus. On full investigation, which included repeated stool examinations, roentgenography of the whole gastro-intestinal tract and repeated sigmoidoscopic examinations, and taking into account the patient's psychological background, it was decided that this was a case of chronic non-specific ulcerative colitis, the precipitating factor being an unhappy life situation.

The patient stated that at the age of 7 years she was confined to bed for several months (without medical attention) for an illness of which she could not recollect the details. On recovery from this illness she had difficulty in walking and over the years she developed a peculiar gait. Before this she had walked normally but had always been a thin and ailing child and often suffered from pain in the limbs. Her left leg became shorter than the right, causing a pronounced limp, and the muscles of the lower limbs were weak. As a result of her disablement she left school at the age of 14 and has never done remunerative work, but has been drawing a government disablement pension since the age of 18 years. About 6 months before the onset of the diarrhoea the pension authorities sent her to a large provincial hospital to ascertain the nature of her disability. An X-ray survey of her skeleton revealed 'a peculiar bone disease' (the patient's own words). The patient further stated that she had never menstruated and had no breast development, and that there was complete absence of libido or interest in the opposite sex. As a result of her disabilities she had the feeling that she was 'different to other women' and was constantly being stared at. She has always been thin and although she has a good appetite she has never been able to gain in weight even before the onset of the diarrhoea. In June 1955 her teeth were extracted in the hope that this would improve the diarrhoea. She is one of 10 children and all the others are in good health. There is no history of a similar ailment in the family.

On examination she was found to be tall (5 feet 9 inches) and very thin (Fig. 1). She walked with a waddling gait and a distinct limp to the left. She appeared to have long limbs, the thighs and forearms being especially long. The forehead was high and wide and the scalp hair fine and blond. There was a slight staring appearance of the eyes but no exophthalmos. The fields of vision and the ocular fundi were normal. The facial skin was smooth and soft and there was slight brownish pigmentation of the forehead, cheeks and bridge of the nose. The conjunctival and oral mucous membranes were pale. The pubic and axillary hair was scanty. The heart and lungs were normal on clinical examination. The blood pressure was 115/70 mm. Hg. There was marked tenderness over the whole of the colon, especially the descending colon, which was easily palpable.

* Formerly Department of Radiology, Pretoria General Hospital, University of Pretoria.

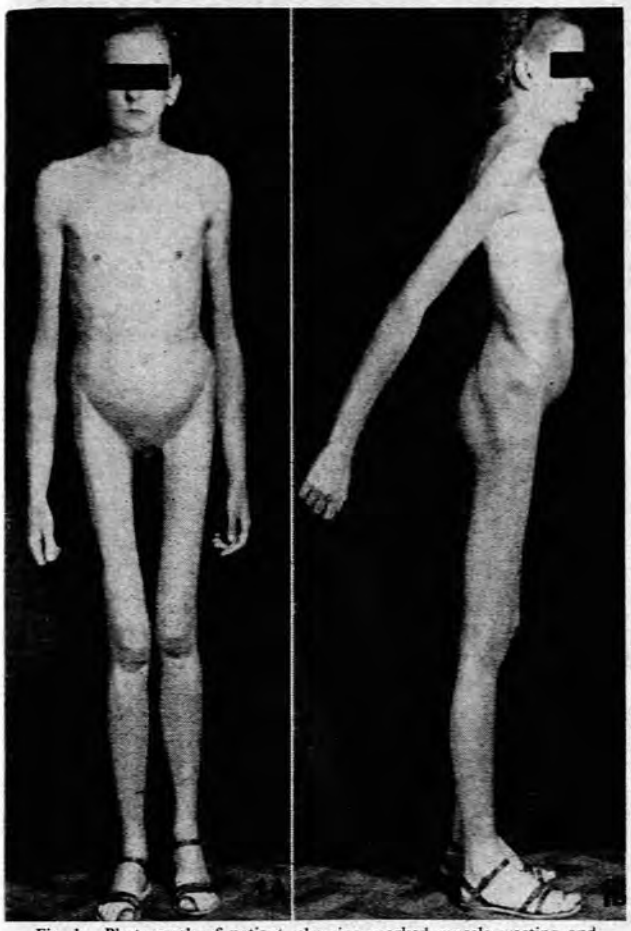

Fig. 1. Photograph of patient, showing marked muscle wasting and absence of breast development.

The breasts were not developed at all. Except for the scantiness of the pubic hair the vulva was normal. The vagina was short and a very small uterus was palpable on vaginal examination. A mass about 1 inch in diameter was felt in the left adnexal area, probably an ovary.

The bones of the arms, forearms, thighs and legs felt thickened. The limb musculature was poorly developed. There was marked limitations of abduction at both hip joints but no limitation in flexion or extension. There was no limitation of movement at the other joints. The tendon reflexes were normal and no disturbance of sensation or other neurological abnormality was found.

Measurements. Length 69 inches, weight 90 lb., pubic height 36 inches, span 73 inches, chest circumference at nipple line 28½ inches, circumference at hips 28 inches, skull diameter 23 inches.

Laboratory findings. Urine analysis was normal. A blood count on 16 March 1955 revealed a severe hypochromic anaemia: Hb. 7·4 g. %, r.b.c. 2,900,000 per c.mm., w.b.c. 5,200 per c.mm. (44% polymorphonuclear leucocytes and 55% lymphocytes). Platelets present in normal numbers. Intramuscular iron therapy (Imferon) was followed by a dramatic improvement in the anaemia; barely 3 weeks after treatment was begun the Hb. rose to 12 g.% and the r.b.c. to 4,000,000 per c.mm., indicating that there was no bone-marrow disturbance causing the anaemia. The modified Ide test yielded a negative result. Chemical analysis yielded the following results: Serum calcium 5·8 mEq./litre; serum sodium 139 mEq./litre; serum potasium 4·1 mEq./litre; blood urea 21 mg./100 ml.; serum albumin 2·3 g.%; serum globulin 2·8 g.%, alkaline phosphatase 13·9 K.A. units. The liver function battery of tests showed equivocal results for liver damage. The sugar tolerance test yielded a normal result. Estimation of 17-ketosteroids in 24-hour urine specimen yielded 3·3 mg. and 2·2 mg.

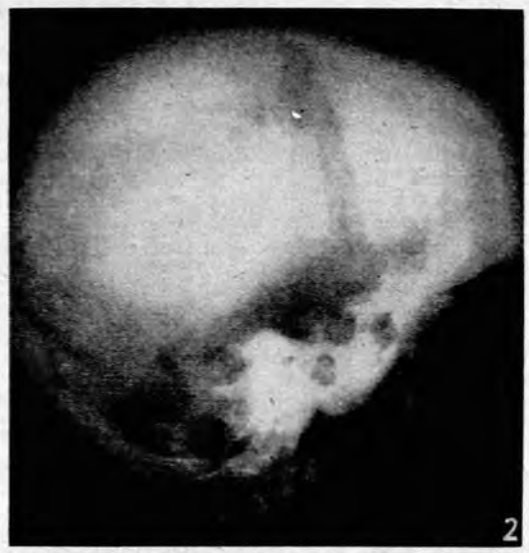

Fig. 2. The skull showing sclerosis of vault and base and involvement of parietal bones.

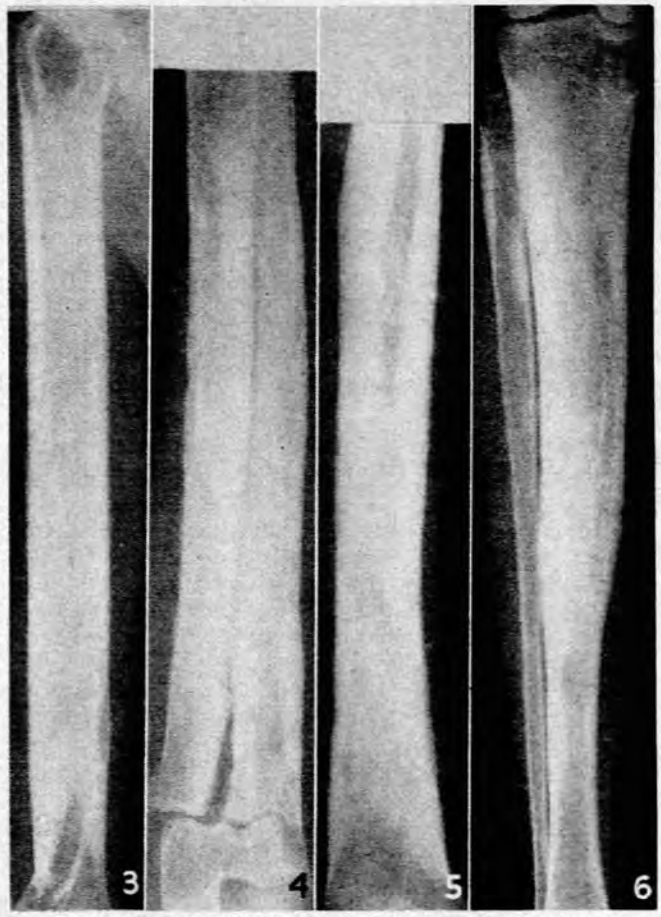

Fig. 3. The humerus showing sclerosis of the shaft and non-involvement of the metaphyses. The medullary cavity is relatively normal. The other humerus shows the same changes.

Fig. 4. Radius and ulna, demonstrating the sclerosis of the shafts. The other radius and ulna show the same changes.

Fig. 5. The femur showing the sclerosis and thickening of the diaphysis. The other femur shows the same changes.

Fig. 6. The tibia and fibula showing the typical changes. The other tibia and fibula show the same changes.

on separate occasions, and 2·1 mg. of 17-hydroxyethiosteroid (estimated as free cortisone). Estimation of follicle-stimulating hormone yielded more than 6 and less than 12 mouse units in a 24-hour urine specimen. Histological examination of a specimen of skin revealed a female chromosomal pattern.

Radiological Examination

There was gross thickening of the vault of the skull, including the parietal bones and the base of the skull; the squamous temporal bones, basi-occiput and sella were normal (Fig. 2). The facial bones and mandible showed no bony changes.

There was symmetrical expansion of the shafts of the humeri, showing gross cortical thickening. The lower ends of the humeri showed no involvement and the metaphyseal and epiphyseal regions were normal. The medullary cavities were relatively uninvolved (Fig. 3). Similar appearances were seen affecting both radii and ulnae (Fig. 4).

Symmetrical and cylindrical expansion was also seen in both femora (Fig. 5). At the metaphyseal region of the femoral necks a dense sclerotic band was present. The femoral heads, however, were normal (Fig. 7). The upper two-thirds of the fibulae and tibiae were similarly affected, leaving the distal thirds quite normal in appearance (Fig. 7). The spine, pelvis, clavicles, hands and feet showed no bony changes.

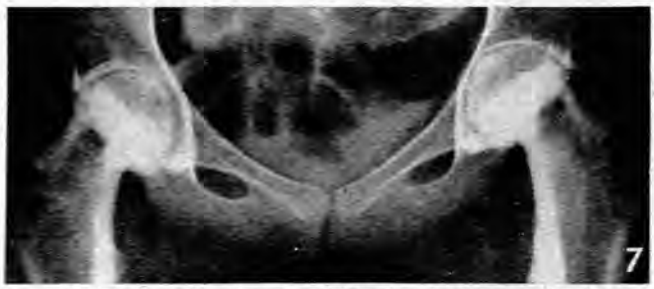

Fig. 7. The femoral heads are normal, but a dense band is seen at junction of head and neck of each femur.

Changes in the femoral necks have only once been reported, by Griffiths.[3] No other case showing the narrow sclerotic band at the junction of the head and neck of the femur could be traced. This case also differs from previously reported cases in that the parietal bones of the skull are also affected.

A barium enema confirmed the presence of ulcerative colitis.

DISCUSSION

Engelmann's disease is characterized by symmetrical enlargement and sclerosis involving the shafts of the major long bones, associated in most cases with skull changes. The changes in the long bones are restricted to the diaphysis, usually the middle two-fourths. According to Griffiths[3] there were skull changes in all but 8 of the 22 cases reviewed by him, with an increased density of the base or vault or both, the changes in the vault involving the frontal bones mainly. Changes were not reported in the parietal bones or the squamous parts of the temporal bones, nor in the facial bones and jaws.[3]

The long bones most often affected are the femora, tibiae, humeri, radii, ulnae, and fibulae. The femora were involved in all cases reviewed by Griffiths[3] and it was these that exhibited the most striking changes. The tibiae were affected in all but 2 of the reported cases. If the tibiae were affected then the fibulae were generally affected as well. The humeri and forearm bones were also involved in most cases.[3] The spine has been reported to be affected in only 1 case,[5] (and then only the atlas was involved), the pelvis in 1 case,[5] the clavicles in 5 cases,[3] and the ribs in 1 case. No other bones have been reported to be involved.

The sex incidence of the disease is about equal, and no case has been described in a negro or in a patient from the Asiatic mainland.[3] The youngest age at which the diagnosis was made was 33 months,[6] and the oldest 55 years.[1] In all adult cases puberty was delayed, the genitalia and secondary sexual characteristics never being well developed. One male adult described as a case of the disease by Stronge and McDowell[7] was sexually normal. Small atrophic testicles have been described in some cases.

Affected children are late walkers and most cases complain of pain in the bones. Most patients are poorly developed and short of stature, the limbs appearing abnormally long in proportion to the height. Postural defects are common and in all cases the gait is described as abnormal and generally of the waddling type. In some cases the abnormality in gait commenced in later childhood. Muscle development is remarkably poor in most cases and the thickened long bones may be palpated in some cases through the thin muscle masses of the extremities. Limitation of hip and knee movements may be present but generally joint movement is free. Investigation of the blood count and blood chemistry have shown no specific or significant deviations. The histology of the involved bone reveals a non-specific picture of osteosclerosis. No evidence of inflammatory changes have been found.

The case described here is presented as one of Engelmann's disease on clinical and radiological grounds. The case presents as a physically underdeveloped female with marked muscle weakness of the extremities, marked muscle wasting, abnormal gait, failure of development of secondary sexual characteristics, and thickened and sclerotic long bones. Radiologically the long bones show the characteristic diaphyseal hyperostosis and the skull the basal and frontal sclerosis. The patient differs from most cases in being tall, only Stronge and McDowell's case[7] also being tall. It is felt that the diarrhoea has nothing to do with the bone condition, which was discovered before to the onset of the diarrhoea. Only in one case, presented for diagnosis by Cockayne[8] in 1920, was looseness of the stools mentioned; Fairbank[9] diagnosed this case as one of Engelmann's disease in later years.

The aetiology of the condition is unknown. Griffiths[3] favours a genetic origin in view of the early onset of the disease, the absence of abnormal cell structure, and the symmetrical progress in the skeleton. It is interesting that Cockayne,[8] even before his case had been given a specific diagnosis, entertained the possibility of an endocrine disorder as being the underlying cause. In view of the late onset of puberty and menstruation in female cases, the presence of small testicles in some of the males, and failure in full development of secondary sexual characteristics in most cases, it is felt that an endocrine disturbance is a possibility as a cause for this condition. It is to be noted that Engelmann's disease is not only a bone disorder but a constitutional disease as characterized by disturbance in general physical and sexual development.

SUMMARY

A case of Engelmann's disease (progressive diaphyseal dysplasia) is presented, with a brief survey of the clinical and radiological features.

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