ENGELMANN'S DISEASE

A 45-YEAR FOLLOW-UP

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We report a 45-year follow-up of a patient with Engelmann's disease previously described in 1950, showing progression of the disease with unique involvement of the femoral capital epiphyses. The case is compared with others to add some information about the later stages of a disease which is not fully understood

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CASE REPORT

This patient was initially described in 1950 by Stronge and McDowell when he was 28 years of age. Over the last 45 years there has been little change in physical appearance, apart from ageing. He is thin and tall with generalised underdevelopment and weakness of the muscles, particularly around the pelvic girdle and thighs. His legs are bowed

Table I. Details of the radiological changes in the skull, spine and arm

	Skull	Spine	Humeri	Radii and ulnae
1950	Sclerotic patches in base and vault maximal in frontal and occipital bone	Normal	Diaphyseal widening Cortical sclerosis Medullary narrowing	Widening, sclerosis Medullary narrowing in upper two-thirds of diaphyses only
Changes to present day	Marked thickening of vault with loss of inner/outer table differentiation	Sclerosis involving posterior arches and posterior fifth of vertebral bodies	Further sclerosis, expansion and bowing Spread to metaphyses Medullary narrowing	Further sclerosis and expansion with involvement of proximal metaphyses

Table II. Details of the radiological changes in the legs, hands and pelvis

	Femora	Tibiae and fibulae	Hands	Pelvis
1950	Widening, sclerosis Medullary narrowing of diaphyses	Widening, sclerosis Medullary narrowing of diaphyses upper two-thirds of diaphyses only	Normal	Normal
1979	Further widening and sclerosis extending into femoral heads and distal metaphyses	Involvement of whole diaphyses with spread into metaphyses		Sclerotic islands in both iliac bones
1986		No change	Cortical thickening of metacarpals of thumbs, right index and ring fingers	
1995	Extension to articular surfaces of femoral heads, seen on CT and MRI	Sclerotic island in right proximal tibial epiphysis		Further iliac sclerosis with involvement of acetabular roofs

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and the lumbar lordosis has increased. His gait is waddling. There is no clinical evidence of involvement of the skull or mandible. His testes are of normal size and he has developed mild splenomegaly although he is not clinically anaemic.

He has always had some pain in his legs after walking short distances, and over the last ten years has also devel-

Fig. 1

Fig. 2

Figure 1 – Radiograph of the left humerus showing cortical sclerosis extending into the humeral head and involvement of the glenoid. Figure 2 – Radiograph of both knees showing cortical sclerosis extending into the metaphyses bilaterally and into the right tibial epiphysis.

Fig. 3

Fig. 4

Figure 3 – Lateral radiograph of the lumbar spine showing sclerosis involving the posterior elements and posterior fifth of the vertebral bodies. Figure 4 – Axial CT of a lumbar vertebral body showing the sclerosis more clearly.

oped pain in the lower back and both knees. He has no visual or auditory symptoms. He has no children and his parents had died before they could be screened for the condition.

There has been a striking radiological progression of the disease, shown in Tables I and II, but serum alkaline phosphatase levels have remained normal. In 1950 the disease involved only the diaphyses of the affected limbs, but it now

Fig. 5

Radiograph of the pelvis showing subarticular involvement of the femoral heads and involvement of the roofs of the acetabula and the iliac bones.

affects the metaphyses of all limbs, the epiphyses and articular surfaces of the femoral heads and acetabula and the right tibial epiphysis. The spine and hands, unaffected in 1950, now show changes (Figs 1 to 6), and there has been some progress of the disease in the skull.

CT and MRI have confirmed the involvement of the articular surfaces of the hips and MRI has clearly shown that the disease involves the cortices, with sparing of the medullary cavities.

DISCUSSION

Engelmann's disease is primarily a disorder of intramembranous ossification and may affect the cortex of long

bones, the skull vault, the midportion of the clavicle and more rarely, the facial bones and mandible, which are all formed by this process. Neuhauser et al (1948), however, described abnormal endochondral ossification in two patients with involvement of the skull base as well as the vault. Although it was first described as a diaphyseal disorder, Naveh et al (1984) found progression of the disease into the metaphyses and we now report late involvement of the epiphyses.

Three grades of clinical severity range from the asymptomatic patient with a radiological diagnosis to the most severe form which presents in infancy with leg pains, difficulty in walking and an abnormal gait (Lennon, Schechter and Hornabrook 1961). Weakness and muscle underdevelopment affect mainly the pelvic girdle and legs and cause a waddling gait. There may also be growth retardation, dry skin with reduced subcutaneous fat, flat feet, valgus ankle deformity, altered reflexes, exophthalmos, nystagmus, auditory impairment and splenomegaly (Lennon et al 1961; Wolf and Ford 1971; Sparkes and Graham 1972; Naveh et al 1984; Greenspan 1991).

The age of presentation ranges from the first to the eighth decade depending on the clinical subtype, but patients usually have a positive family history, showing an autosomal dominant mode of inheritance. The existence of sporadic forms is disputed, since other asymptomatic family members may not have been screened radiologically (Sparkes and Graham 1972).

Our patient shows typical radiological appearances in the diaphyses, pelvis and skull, and is the first Caucasian to show spinal involvement confined to the posterior elements and the posterior fifth of the vertebral bodies (Wolf and Ford 1971) (Figs 3 and 4). Metacarpal and metatarsal involvement is rare (Lennon et al 1961); we accept that the acrosclerosis may be a normal variant.

The normal alkaline phosphatase in Engelmann's disease differentiates it from Van Buchem's disease and chronic

Fig. 6

Axial CT of the pelvis showing subarticular sclerosis in the femoral heads.

hyperphosphataemia (Lennon et al 1961; Iancu et al 1978; Greenspan 1991). In our case, the early age of onset, the symmetry of bone involvement and the clinical symptoms differentiate it from Ribbing's disease (Greenspan 1991) while the main finding in autosomal dominant sclerosis is mandibular involvement (Gelman 1977).

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