A. E. A. RIDGWAY: SOLAR RETINOPATHY

FIG. 1.—Case 3. Photograph taken on 24 May. Kodachrome II. Kowa fundus camera.

FIG. 2.—Case 5. Photograph taken on 3 June. Kodachrome II. Nordens fundus camera.

P. D. GULATI *ET AL.*:
ENGLEMAN'S DISEASE

FIG. 1

FIG. 2

FIG. 3

FIG. 1.—Radiograph showing increased density of the diaphysis of long bones of the lower extremity. Epiphysial and metaphysial zones show normal density.

FIG. 2.—Radiograph showing increased density of the diaphysial zones of long bones of the upper extremity.

FIG. 3.—Radiograph of skull showing osteosclerosis of base of skull as well as bones of calvarium.

Medical Memoranda

Case of Englemann's Disease

[WITH SPECIAL PLATE]

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Englemann's disease is one of the rarest bone dystrophies causing osteosclerosis. The first case was published by Englemann in 1929. A search of the literature has revealed 53 cases reported up to 1964. However, there has hardly been any record of a similar case in the Indian literature. The purpose of this paper is to report a case of Englemann's disease and to review the literature on the subject.

CASE REPORT

A girl aged 14 was admitted to our medical unit of Irwin Hospital, New Delhi, with the object of investigating the cause of her stunted growth. Her father had noticed that her growth had not been up to the mark since the age of 4 years. She had had slight pain in the legs for one year. Her developmental milestones were normal except that she started walking at the age of 2 years. Past and family history were non-contributory. The outstanding features on general examination were a short stature, a waddling gait, and the absence of secondary sex characters. She had not started menstruation and her genitalia appeared infantile. Her height and span were below average for her age and her crown-to-rump and rump-to-heel ratio was 1.2:1. Local examination revealed thickening, tenderness, and venous prominence over both tibial bones. The ulna was irregularly palpable on both sides. Examination of nervous, cardiovascular, respiratory, and gastrointestinal systems showed nothing abnormal.

Investigations.—Haemoglobin was 12.0 g./100 ml., total leucocyte count 7,700/cu. mm. (polymorphs 50%, lymphocytes 49%, eosinophils 1%), erythrocyte sedimentation rate 9 mm. in first hour (Westergren). Urine and stool examinations showed nothing abnormal. Serum alkaline phosphatase was 4.1 K.A. units, serum calcium 11.2 mg., serum phosphorus 3.7 mg., blood urea 23 mg., and serum cholesterol 150 mg./100 ml., glucose-tolerance curve normal. Twenty-four-hour excretion of fat in stool was 3.5 g., and 24-hour excretion of 17-ketosteroids in urine was 5.9 and 3.2 mg./100 ml. Basal metabolic rate was +8. Mantoux test was positive. Buccal smears for sex chromosomes was positive (female type). Radiological changes were seen in the diaphysis of all the long bones as well as in the skull bones. Bones of hands, feet, clavicle, ribs, spine, scapulae, and pelvis did not show any changes. The typical findings were in the form of cylindrical expansion of the whole of the diaphysis due to cortical thickening which reduced the width of the medullary canal (Special Plate, Figs. 1 and 2). The metaphysis and epiphysis were left unaffected. The skull showed gross thickening and sclerosis of the frontal bones, particularly the supraorbital margins, extending towards the forehead as well as to the floor of anterior and middle cranial fossae and backwards to the petrosal ridge. The parietal and frontal bones showed patchy areas of sclerosis (Special Plate, Fig. 3). Biopsy from the affected bone showed moderate thickening of the bony trabeculae with normal laying down of osteoblasts.

COMMENTS

Englemann's disease is characterized by underdevelopment due to fusiform expansion and sclerosis of the diaphysis of long bones. The disease is diagnosable in infancy and may be suspected in any child with feeding problems and delayed walking. The youngest patient reported by Gvozdanovic (1950) was a 3-months-old girl. During childhood aching limbs and a wide-based waddling gait are characteristic features. Delayed puberty

associated with or without skeletal deformities and a variable degree of neurological deficit are common in later life (Riley and Shwachman, 1943). The disease, as in the present case, is often discovered accidentally.

A multitude of laboratory tests revealed no abnormality. However, radiological changes are diagnostic. Classical changes start in the diaphysis of long tubular bones with spindle-shaped sclerotic thickening, and as the age advances osteosclerosis extends lengthwise but the metaphysial and epiphysial zones are usually not affected, though Clawson and Loop (1964) described an unusual case showing widening and osteoporosis of these areas as well. The characteristic changes have been seen mostly in the long bones of extremities, but others are also involved. However, Lennon *et al.* (1961) in a review of 52 cases described the involvement of the base of the skull in 53%, cervical vertebrae in 9.6%, and metatarsals and metacarpals in 9.6%. Long bones as well as skull bones are involved in our case.

The histological picture is very variable (Neuhauser *et al.*, 1948; Bingold, 1950). The changes, whenever present, are confined to the radiologically altered areas of bone and consist mainly of cortical hyperostosis. Histologically, the disease has to be differentiated from infantile cortical hyperostosis, which occurs in infants and where the involvement of bone is asymmetrical and ribs and mandible are invariably involved (Caffey, 1956; Stewart and Cole, 1956).

The exact pathogenesis of the condition is not known. Englemann (1929) believed that it was caused by a disturbance of the haemopoietic system, but this belief has not been upheld by subsequent workers (Neuhauser *et al.*, 1948; Sear, 1948; Bingold, 1950; Griffiths, 1956). Some suggestion of pituitary deficiency mainly based on the occurrence of infantilism associated with diminished excretion of 17-ketosteroids in urine has been made by Jammes *et al.* (1953) and Buchem *et al.* (1955). However, in our case urinary 17-ketosteroid excretion was within normal limits. The disease is often reported to be familial, though the genetic basis has not been fully worked out.

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