Case report 492*

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Radiological studies

Fig. 1. A roentgenogram of the innominate bones shows alterations in density of bone with translucent areas and areas of increased density predominantly affecting the femoral metaphyses. In addition, observe the area of expansion of bone in the right ischium and right inferior pubic ramus.

Clinical information

Tibial osteotomies were performed at the ages of three and eight years in an attempt to straighten the legs of this male child.

At the age of 28 years he presented with visual field defects and a basophil adenoma was removed from the pituitary gland.

He was noted at this time to have the classical radiological and clinical features of a skeletal dysplasia (to be named later) with marked deformities of the hands and feet.

In spite of severe dwarving and considerable disability the patient lived a normal life, working as a proofreader and only attending hospital for renewal of surgical shoes.

At the age of 30 years the index and the little finger of his right hand were so grotesquely enlarged that he requested surgery to improve their function and appearance. A 7 cm diameter mass was removed from each finger and he was so pleased with the result that 2 years later at his request similar surgery was performed on the left ring finger.

Five years later at the age of 35 years he presented with a one-year history of progressive pain in the lower lumbar region radiating to the right...
Fig. 2. A An isotope bone scan shows increased uptake over the area of the right ischium. B An isotope bone scan shows increased uptake in the left hand and clavicle.

groin. The pain was made worse by lying down and prevented sleep. His job was also prejudiced since he could sit only with difficulty. On examination tenderness was elicited at the right ischial tuberosity and a tender mass was palpated in the right buttock. No neurological abnormality was discernable and mobility of the spine above the hips was normal.

An AP roentgenogram of the pelvis (Fig. 1) showed multiple areas of altered density of bone and deformity of the pelvic bones with areas of cortical expansion. An area of cortical destruction with amorphous calcification was noted in the right ischium. An isotope bone scan showed markedly increased uptake in the region of the right ischium (Fig. 2A). Increased uptake was also observed in the hand (Figure 2B).

An open biopsy of the right ischium was performed.