GAUCHER'S DISEASE WITH PSEUDOCOXALGIA

Report of a Case


An undersized boy aged four years and ten months was admitted to King's College Hospital in 1946 under Dr Phillip Evans because of failure to gain in weight. There was a past history of frequent colds and malaise. On examination the spleen and liver were found enlarged. There was generalised glandular enlargement. Radiographs of the skull, chest and long bones were normal. Wassermann reaction was negative. Blood cholesterol was 174 milligrams per cent. A tentative diagnosis of Gaucher's disease was made.

Progress—Two years later, at the age of six years, he was readmitted because of pain and limp in the right leg for three months. On examination there was no wasting of the thigh muscles. Flexion of the right hip was full, but rotation and abduction were limited. Radiographs showed a cycle of epiphysial changes in the head of the right femur (Figs. 1 to 6). Investigations—Gaucher cells were present in sections of bone marrow. Treatment—Extension was applied in abduction for three months, after which a patten-ended caliper was worn.
Figure 3—Three months later a cyst is now visible in the metaphysis. Figure 4—Three months later still. The epiphysis is fragmented and two large cysts are visible in the metaphysis.

Figure 5—One year later. Reformation of ossific centre. Figure 6—Recent film, three years after the first (Fig. 1) showing coxa magna. A cystic area is still visible in metaphysis.

Comment—This case of Gaucher's disease is so far devoid of osseous lesions other than those in the right hip joint. The serial radiographs (Figs. 1 to 6) differ from those seen in typical pseudocoxalgia only in the lack of initial condensation of the epiphysis. Arkin and Schein (1948) have drawn attention to the fact that such changes may be brought about by deposits of Gaucher cells in the femoral neck or head.

Reference