GAUCHER’S DISEASE WITH INVOLVEMENT OF BOTH HIP JOINTS

Report of a Case

O. J. VAUGHAN-JACKSON, LONDON, ENGLAND

The patient, a clerk aged fifty-four years, attended St Bartholomew’s Hospital, Rochester, in 1950 complaining of pain in both hips. At the age of twelve years he had had what was thought to be a tuberculous left hip, but he was never treated in bed but only with a Thomas’s caliper and crutches. With this treatment his hip recovered completely, but since about 1916 there had been a slow deterioration in the hip both in comfort and mobility. Since that time the right hip also had gradually become stiff and painful. In 1936, when he was aged forty years, an arthroplasty of the right hip was carried out by Mr Laming Evans at the Royal National Orthopaedic Hospital. After this the hip was comfortable and during the recent war he carried out active duties in air raid defence. Since 1945 the right hip had been getting painful again, and he was now only able to walk about half a mile before pain forced him to stop. The left hip was also painful but to a lesser degree. He had had some pain in the low back after walking. In addition to the operation on the right hip, he had loose bodies removed from the left knee in 1934 and 1941.

On examination he appeared healthy but rather sallow. There were marked triangular subconjunctival lipoid deposits in both eyes. He had a marked lumbar lordosis, secondary to a 70 degrees flexion deformity of the right hip. In the left hip there was a 30 degrees flexion deformity. There was only a degree or so of movement in any direction in the right hip. In the left hip he had a range of active and passive flexion from 150 degrees to 110 degrees. All other movements of the hip were restricted to a few degrees. In the left knee there was movement from 160 degrees to 80 degrees, with crepitus and clicking in the joint and marked lateral instability. The right knee showed no abnormality. On walking and standing there was a bilateral Trendelenberg dip. He could touch his toes, but on resuming the erect position there was a very loud crunch in the right hip. Lateral movements of the spine were free. Abdomen—The spleen showed simple smooth enlargement to below the umbilicus. The liver was enlarged three fingers’ breadths below the costal margin. Radiographs showed gross

Fig. 1
Pelvis and hip joints.
arthritis of the left hip with flattening of the femoral head, and gross changes in the structure of the right hip, the head of the femur being absent (Fig. 1). In both hips there were widespread patchy sclerotic changes throughout the upper ends of the femora and adjacent parts of the ilium. In the upper part of the left femur there was an area in which the normal bone pattern was lost. The lower end of each femur showed a “bottle” enlargement with widespread alteration of bony pattern (Figs. 2 and 3). The upper ends of both tibiae showed similar alterations of pattern. The left knee was the site of marked arthritic changes (Fig. 1). Radiographs of the skull, arm and forearm bones showed no evidence of additional deposits, and the lumbar spine showed very little arthritic change.

Investigations—Blood examination showed a mild leuco-erythroblastic anaemia with a red cell count of 3,320,000; haemoglobin was 66 per cent. One per cent of metamyelocytes and two late normoblasts were seen during a differential white cell count of 200 cells. Sternal puncture showed marked normoblastic hyperplasia. Gaucher’s cells were not seen. The bromsulphthalein test showed no retention of dye after thirty minutes. Despite the absence of clinching evidence of Gaucher’s disease, the diagnosis did not seem in doubt.

Progress—The patient was admitted to the London Hospital three months later for further investigations. The acid phosphatase was found to be raised to 21 units (this was repeatedly checked) and the blood cholesterol was 140 milligrams per cent. Sternal puncture revealed Gaucher’s cells present in small numbers. It was noticed that when a sphygmomanometer cuff was applied petechiae appeared over the arm and forearm below the cuff. Further blood investigations were carried out, and these revealed a bleeding time of eight and a quarter minutes and the blood platelet count was reduced to 20,000. The question of treatment for the hips is under consideration.