Acta Orthop Scand 57, 171–172, 1986

Hip arthroplasty for alkaptonuric ochronosis

A case report

Ochronotic arthropathy is present in about one third of the patients with alkaptonuria. The large joints, as well as the spinal column, are affected. In a typical case with a grave classic ochronotic arthro- and spondylopathy, arthroplasties of the hips greatly increased the patient's physical activity.

Correspondence: Tölö Hospital, Topeliuksenkatu 5, SF-00260, Helsinki 26, Finland

Antero Mäkelä
Olli Korkala

Department of Orthopedics and Traumatology, University Central Hospital, Helsinki, Finland

Alkaptonuria (ochronosis) is a rare metabolic disease, which is inherited as a recessive Mendelian trait. It is characterized by excretion of homogentisic acid in the urine, due to the absence of homogentisic acid oxidase in the liver and kidneys (Zannoni et al. 1962). Homogentisic acid has a special affinity for cartilage, which becomes brittle and disintegrates. In about one third of the cases, a secondary degenerative arthritis affects the large joints, whereas the smaller joints are involved only to a minor degree (Kolar & Krizek 1968, Mohr 1983).

We performed arthroplasties on both hips in a grave case of ochronosis.

Case report

A 57-year-old man had a history of low-back and neck pain since the age of 30. He was first seen at our hospital in 1972. He had stopped his heavy work three years earlier, but his symptoms had still gotten worse. On admission in 1972, his general condition was good, but he looked much older than his age. He was bowlegged and had a fixed kyphosis posture of the thoracolumbar spine, atrophy of the shoulders and upper extremity musculature, and effusions of both knees. He also complained of grave pain in the thighs, knees, and back, and he had developed a very careful and slow walking pattern. There was a slight bluish pigmentation in the auricles and scleras.

He had severe radiographic changes in the shoulder and knee joints, whereas the hips and sacroiliac joints were affected to a somewhat lesser degree (Figure 1). Severe disk narrowings and disk calcifications were noticed, as well as symphyseal and auricular ossifications. These findings aroused our suspicion that ochronosis was present.

Blood cell counts and morphology were within normal limits. Serum electrophoresis was normal, but the liquor immunoelectrophoresis showed all the components of the serum protein fractions and appeared to be pathologic. The albumin/IgG ratio was normal. Urine sedimentation was normal. Homogentisic acid was present in the urine, but koproporphyrins and uroporphyrins were lacking. Electroence-

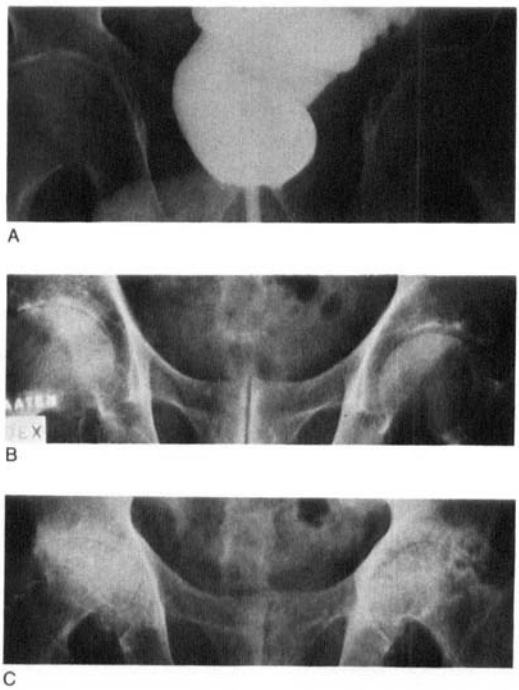

Figure 1. Progress of ochronotic hip arthropathy. A 1974. Normal hips in conjunction with a barium enema. B 1979 and C 1984 show increasing arthropathy.

phalography and electroneuromyography of the peripheral muscles showed normal features. Muscle histology was also normal.

The diagnosis of ochronosis was therefore made. The urine of the patient's four children was analyzed for homogentisic acid, which was not detected.

In 1984, the patient returned with intolerable pain in his hips and back. He was now moving about slowly with the aid of two crutches. He had flexion contracture with very poor movements of both hips. Radiographically, the arthropathy of the hips had progressed markedly (Figure 2). Hip arthroplasty, resulting in a marked improvement in his condition, was performed first on the left side in August 1984 and then 8 months later on the right side. During the operations of the hip joints, some residual black strips of cartilage remnants were found; however, mostly bare subchondral bone was seen at the articular surfaces on both sides. The histologic examination of the femoral heads showed cystic degeneration of the subchondral bone and the presence of black pigment in both the cartilage and the bone. The patient was succesfully rehabilitated to an independent life at home. At his last visit in August 1985, he still used crutches, but not because of his hips, which were now painless and for the first time in many years he could ride a bicycle without experiencing any pain. His painful lower back was now the cause of his using crutches.

Discussion

There is no cure for ochronotic arthropathy. Reduction in the intake of food containing tyrosine and phenylalanine and the administration of high doses of vitamin C reduce the excretion of homogentisic acid, but they have no effect on the progress of the disease (Turek 1984). Physiotherapy of the back and affected joints plays an important role. Modern orthopedic surgery using artificial joints increases the chances of active rehabilitation of patients with ochronotic arthropathy.

References

Cervenansky, J., Sitaj, S. & Urbanek, T. (1959) Alcaptonuria and ochronosis. *J. Bone Joint Surg.* **41-A**, 1169–1182.

Harrold, A. J. (1956) Alcaptonuric arthritis. *J. Bone Joint Surg.* **38-B**, 532–538.

Kolar, J. & Krizek, V. (1968) Alkaptonurische Ochronose. *Med. Klin.* **63**, 1105–1108.

Mohr, W. (1983) Pathogenese und Morhologie progressiver Gelenkschäden. *Orthopäde* **12**, 78–96.

Srsen, S., Cisarik, F., Pasztor, L. & Harmecko, L. (1978) Alkaptonuria in the Trencin district of Czechoslovakia. *Am. J. Med. Genet.* **2**, 159–166.

Turek, S. L. (1984) *Orthopaedics – Principles and their application.* 4th ed., pp. 251–253. Lippincott Co., Philadelphia, USA.

Zannoni, V. G., Seemiller, J. E. & La Du, B. N. (1962) Nature of the defect in alcaptonuria. *Nature* **193**, 952–953.