A CASE OF ALKAPTONURIA WITH ROOT CANAL STENOSIS

K B H Koh, E H Low, S L Ch'ng, I Zakiah

ABSTRACT

Spinal involvement in alkaptonuria is common. Patients usually present in the third or fourth decade with spondylosis or acute intervertebral disc prolapse. Alkaptonuria with root canal stenosis has however hitherto not been reported. We wish to report one such patient.

Keywords: *alkaptonuria, root canal stenosis.*

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INTRODUCTION

Alkaptonuria is a rare disorder. About seven hundred cases have been reported in the world literature to date[1]. The disorder is due to a deficiency of homogentisic acid oxidase in the liver and kidney with accumulation of homogentisic acid (an intermediate in the metabolism of phenylalanine and tyrosine) and its products (polymeric ochronotic pigments) in tissues. This contributes to the ochronotic manifestation of the disease. The condition could present insidiously as ochronotic osteoarthritis or spondylosis, or acutely with rupture of the nucleus pulposus. It could be associated with mitral and aortic valve disease, prostatic or renal calculi. We report a case of ochronosis with an unusual presentation of root canal stenosis.

CASE REPORT

A 32-year-old Indian labourer presented with a two-year history of intermittent low backache and right-sided sciatica of one week's duration, associated with numbness and weakness of his right lower limb. There was no sphincteric disturbance. He also noticed that his urine became dark when left to stand.

Physical examination showed a well-developed man with grey pigmented pinnae. He has thoracic kyphosis, loss of lumbar lordosis, and limitation of back movement because of pain. The straight leg raising test was limited at an angle of 60° on the right side and normal on the left. There was weakness of the right extensor hallucis longus muscle and sensory hypoaesthesia on the dorsum of the right foot. The reflexes were normal. Chest expansion was normal and there were no abnormalities in his other joints.

Department of Orthopaedic Surgery
Faculty of Medicine
University of Malaya
Kuala Lumpur
Malaysia

K B H Koh, MBBS, FRCS

E H Low, MBBS, FRCS

Department of Pathology
Faculty of Medicine
University of Malaya

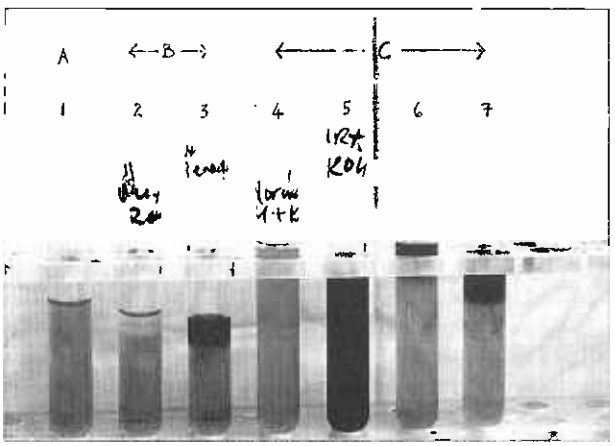
S L Ch'ng, MBBch, FRCPath, FRCPA

I Zakiah, MBBS, MPath

Correspondence to: Dr K B H Koh
Department of Urology
St James' University Hospital
Beckett Street
Leeds LS9 7TF
United Kingdom

Laboratory investigation showed normal haematological parameters and a normal renal function test. His urine became dark brown on standing overnight. Addition of concentrated potassium hydroxide solution accelerated the change and the urine gave a positive reducing reaction with Benedict's reagent. (Fig 1). Urine thin layer chromatography (TLC) on silica gel showed the presence of homogentisic acid and absence of abnormal reducing sugars[2]. Homogentisic acid concentration[3] in three specimens of his urine ranged between 1.09 and 1.14 g/L, while in 8 normal controls it was between 0.026 and 0.160 g/L. A diagnosis of alkaptonuria was thus made.

Fig 1 – (from left to right)
(A) – Patient's urine left to stand (tube 1).
(B) – Reaction of Benedict's solution with normal urine giving a negative reaction (tube 2), and with patient's urine giving a positive reaction (tube 3).
(C) – Reaction with potassium hydroxide with normal urine (tube 4), and with patient's urine (tube 5 left to stand, tubes 6 and 7 reacted immediately).



Radiographs of his spine showed increased thoracic kyphosis and loss of lumbar lordosis. There was widespread narrowing of the intervertebral disc spaces with wafer-like calcification and the presence of the 'vacuum phenomenon' at multiple levels. The intervertebral foramina were narrowed (Fig 2). CT of the spine at L4 to S1 level showed marked degenerative change at the facet joints especially between L5 and S1 vertebrae with prominent osteophytes encroaching into the spinal canal and intervertebral foramina (Fig 3). There was no herniation of the intervertebral disc.

The patient was first treated conservatively with bed rest and indomethacin but this failed to alleviate his symptoms.

Fig 2 – Lateral radiograph of the spine showing narrowing of disc spaces with vacuum phenomenon (arrows)

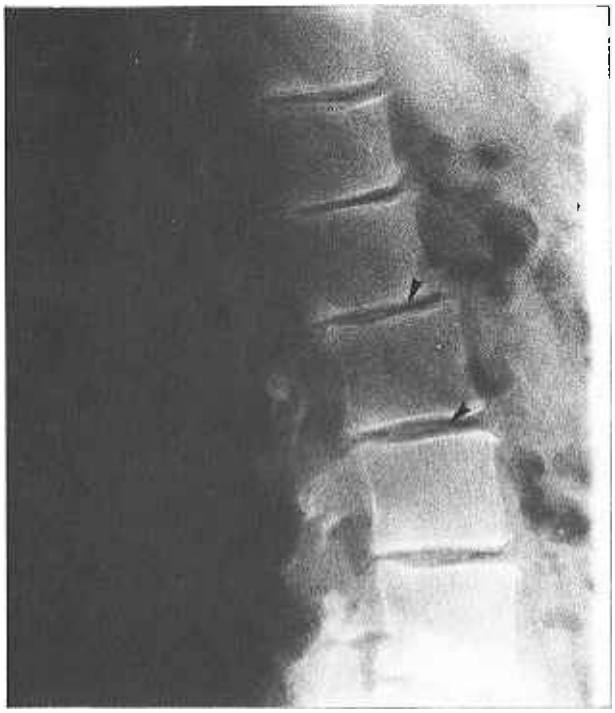
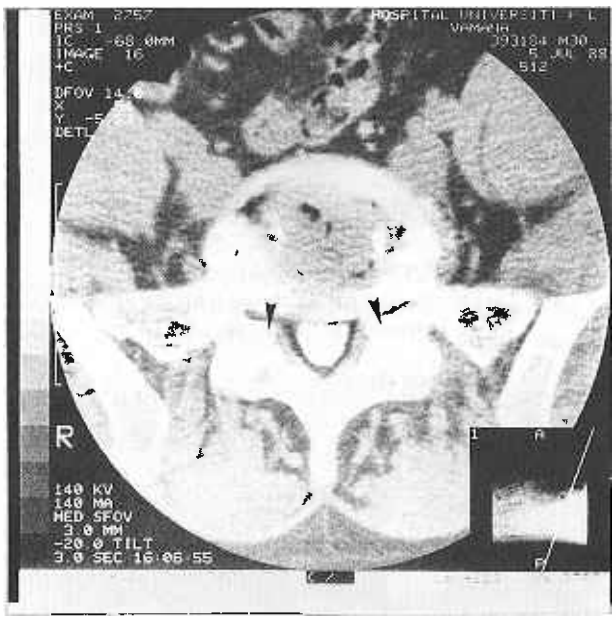


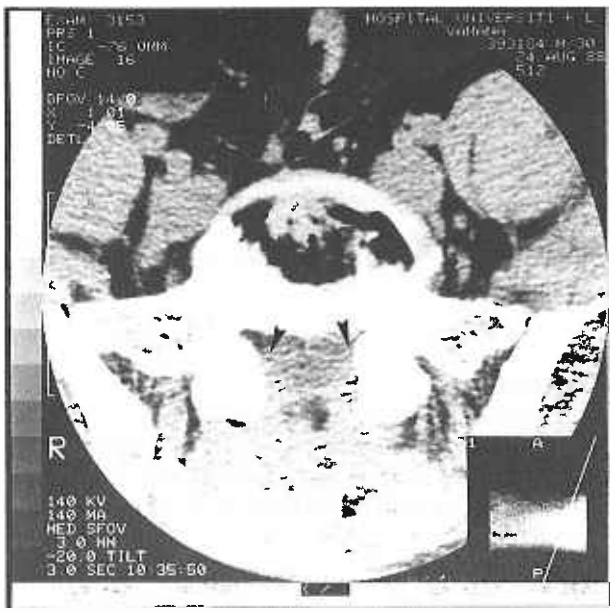
Fig 3 – CT at L5 level showing marked lateral root canal stenosis caused by facet joint osteophytes (arrows).



Subsequently, laminectomy with partial facetectomy to decompress the lateral root canal was performed at L5 level. At surgery, the interspinous ligament was noted to show dark pigmentation. Post-operatively the patient showed a dramatic improvement and his subsequent course was uneventful. Repeat CT showed successfully decompressed root canals (Fig 4).

Examining the patient's family, we found consanguinity between his parents. Thin layer chromatography of urine specimens of his mother, sisters and daughter for homogentisic acid was positive only in his sister.

Fig 4 – Post operative CT scan showing laminectomy with decompressed root canals (arrows).



DISCUSSION

Alkaptonuria is a rare disease and has attracted the interest of clinical scientists since the fifteenth century. The condition has been comprehensively reviewed by O'Brien[4]. Most alkaptonurics present early in the fourth decade with a chronic progressive backache and stiffness as a result of ochronotic spondylosis. This is usually followed by knee, shoulder and hip involvement and the patient becomes increasingly crippled.

This case (the third to be reported from Southeast Asia[5]) shows the typical biochemical abnormalities of alkaptonuria. It is thought that the polymeric ochronotic pigment binds to collagen fibers and alters its chemical structure, and this predisposes the spine to degenerative change[6]. The auto-oxidation of homogentisic acid and generation of free radicals could also contribute to the changes[7].

Facet joint degeneration leading to root canal stenosis is a widely recognised pathological entity. However, on reviewing the literature, we have not found this described in ochronotic spondylosis. Changes in the facet joints in ochronosis that have been described include articular space narrowing, bony sclerosis and even ankylosis[8-10]. In contrast to commoner types of degenerative spinal disorders, vertebral osteophytes in ochronosis are poorly developed[4,11]. We think the florid osteophytosis in this patient is the result of facet joint instability coupled with excessive stresses from the heavy manual work he was engaged in.

References

1. Christensen K, Manthrope R. Alkaptonuria and ochronosis. Hum Hered 1983; 33: 140-4.
2. Ibbott FA. Aminoacids and related substances. In: Henry RJ, Cannon DC, Winkerman JW. eds. Clinical Chemistry – Principles and Techniques. New York: Harper and Row Publishers 1975: 625-6.
3. Neuberger A, Ramington C, Wilson JMC. Studies on alkaptonuria – investigation on a case of human alkaptonuria. Biochem J 1947; 41: 438-49.
4. O'Brien WM, La Du BN, Bunim JJ. Biochemical, pathologic and clinical aspects of alkaptonuria, ochronosis and ochronotic arthropathy. Am J Med 1963; 34: 813-37.
5. Haridas G, Wong HB. Alkaptonuria. J Singapore Paed Soc 1962; 4: 35-8.
6. Zannoni VG, Lomtevas N, Goldfinger S. Oxidation of homogentisic acid to ochronotic pigment in connective tissue. Biochemical et Biophysica Acta 1969; 177: 94-105.
7. Martin JP Jr, Batkoff B. Homogentisic acid autoxidation and oxygen radical arthritis. Free Radic Biol Med 1987; 3: 241-50.
8. Lagier R, Sit'aj S. Vertebral changes in ochronosis. Ann Rheum Dis 1974; 33: 86-92.
9. Pomeranx MM, Friedman LJ, Tunick IS. Roentgen findings in alkaptonuric ochronosis. Radiology 1941; 37: 295-300.
10. Thompson MM. Ochronosis. Am J Roentgen 1957; 78: 46-53.
11. Resnick D. Alkaptonuria. In: Resnick D, Niwayama G. eds. Diagnosis of Bone and Joint Disorders. Philadelphia: WB Saunders Co 1981: 1620-36.