Journal of Clinical Orthopaedics and Trauma 8 (2017) S80–S83

Contents lists available at ScienceDirect

Journal of Clinical Orthopaedics and Trauma

journal homepage: www.elsevier.com/locate/jcot

Case report

Arthroscopic diagnosis and treatment of shoulder ochronotic arthropathy – A case report

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ARTICLE INFO

Article history:
Received 30 July 2016
Received in revised form 5 November 2016
Accepted 17 November 2016
Available online 5 January 2017

Keywords:
Ochronosis
Shoulder
Arthroscopy
Alkaptonuria

ABSTRACT

Alkaptonuria is a rare inherited metabolic disorder, caused by the deficiency of homogentisate 1,2 dioxygenase enzyme. The three major features of alkaptonuria are the presence of homogentisic acid in urine, ochronosis (bluish-black pigmentation in connective tissue) and arthritis of the spine and large joints.

We present a 48 years old female presented with pain, restriction of movements of right shoulder. Arthroscopy was suggestive of ochronotic arthropathy. The definitive diagnosis of ochronosis was subsequently confirmed by laboratory and pathologic evaluation.

1. Introduction

Alkaptonuria is a rare genetic disorder caused by deficiency of homogentisate 1,2-dioxygenease (HGD) leading to failure in processing of amino acids phenylalanine and tyrosine, thus causing accumulation of an intermediate substance homogentisic acid (HGA) in blood and tissues. This HGA and its oxidated form *alkapton* are excreted in urine leading to darkening of urine on long exposure to air. HGA oxidizes to benzoquinone acetic acid, which in turn form melanin like polymer which is deposited in collagen causing ochronosis. This leads to damage to cartilage which causes early joint destruction and painful degenerative arthritis involving knee, spine and shoulder.

We present a patient presented with shoulder pain and stiffness, who was diagnosed as a case of ochronosis on shoulder arthroscopy.

2. Case report

A 48 years old female presented with pain and limitation of movements in the right shoulder for 3–4 months duration which gradually worsened with time. Her activities of daily living were severely affected and she was unable to get sound sleep because of pain. She took analgesics regularly but was unable to get satisfactory relief of symptoms. She also had mild pain in her both knees and left shoulder.

On clinical examination, significant wasting was seen around shoulder. Diffuse tenderness was present all around the shoulder. Active shoulder movements were painful and grossly restricted. Passive range of motions was: abduction of 60°, forward flexion of 80°, external rotation of 10°, and internal rotation up to gluteal area (30°).

Laboratory tests including complete blood counts, ESR, CRP, Serum uric acid were normal and RA factor, HLA B27 were negative. Radiograph of right shoulder showed superior migration of the humeral head, decreased joint space and degenerative changes in the humeral head (Fig. 1). MRI of the right shoulder demonstrated joint effusion, synovial thickening, articular erosion and flattening of the humeral head possibly because of chronic inflammatory process with degenerative changes (Fig. 2).

Non-operative treatment in the form of NSAID and rehabilitation followed by local corticosteroid injection failed to improve the symptoms. Hence it was decided to go ahead with an arthroscopic examination.

Arthroscopy of the right shoulder was performed under general anaesthesia in using standard posterior and anterior portals. Dark brown-black, soot like pigmentation of the synovial tissue and cartilage was noted. Also there was erosion of the glenoid, labrum and humeral head, synovial hyperplasia and osteochondral loose bodies in the joint (Fig. 3). Debridement of the hypertrophied

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http://dx.doi.org/10.1016/j.jcot.2016.11.009
0976-5662/

Fig. 1. Radiograph of right shoulder showing superior migration, decreased joint space and degenerative changes in the humeral head.

Fig. 2. MRI of the right shoulder demonstrated joint effusion, synovial thickening, articular erosion and flattening of the humeral head possibly because of chronic inflammatory/infective process with degenerative changes.

synovium and pathological tissue along with removal of loose bodies was done. A biopsy of synovium and frayed tissue was taken. Postoperatively, the patient was given cuff and collar sling for two days and rehabilitation was started.

Based on arthroscopic findings, provisional diagnosis of alkaptonuria was thought and patient re-evaluated in light of fresh findings. Clinically, she showed blue discolouration of the auricular cartilage (Fig. 4), kyphoscoliosis of the dorsolumbar spine, thickening and tenderness in left Achilles tendon. Radiographs of spine revealed narrowed disc spaces with wafer like horizontal bands of calcification of each space (Fig. 5). The knee joint radiographs showed gross narrowing of the joint space

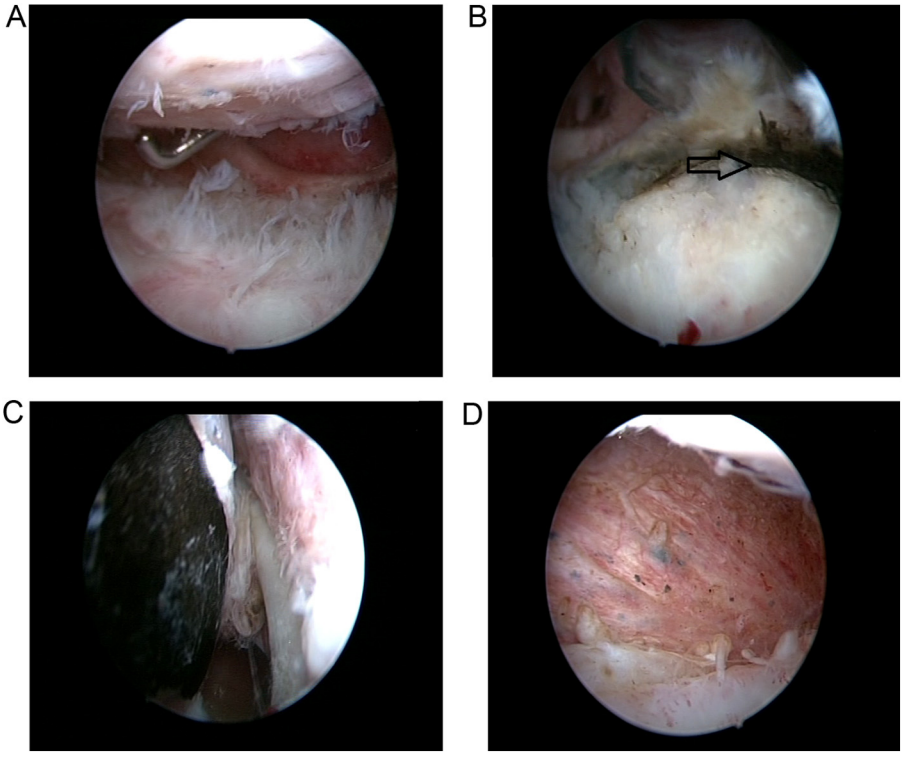

Fig. 3. (A) Arthroscopic view of the right shoulder showing erosion of glenoid and head. (B) Arthroscopic view of the right shoulder showing pigment behind anterior and superior labrum. (C) Arthroscopic view of the right shoulder showing pigment over posterior superior part of head. (D) Arthroscopic view of the right shoulder showing pigmentation of anterior capsule and labrum.