The patient, a 43 year old woman with a 35 year history of alkaptonuria (fig 1A), presented to the orthopaedic clinic for pain in the right shoulder. Clinical examination demonstrated an important reduction of shoulder mobility with, on radiographic evaluation, a greatly destroyed joint (fig 1B). The patient was admitted to hospital to obtain a shoulder prosthesis.

Ochronosis is a rare autosomal recessive disorder featuring total lack of the enzyme homogentisic oxidase, resulting in an accumulation of homogentisic acid, a metabolite of phenylalanine and tyrosine. Deposition of ochronotic pigment results in darkening of the skin and cartilage, which is first seen in the conchae, antihelix of the ear (fig 1C), and sclera.1

Macroscopic inspection of the joint during surgery showed darkening of the cartilage. Histopathological examination of the paraffin embedded sections disclosed brittle pigmented articular cartilage, of which very small fragments were broken off and displaced into the synovial tissue: yellow-brown shards, scattered over the synovium (fig 1D). These typical shards can evoke foreign body reactions with multinucleated giant cells (fig 1E) and induce formation of new bone tissue called osteochondral bodies. Furthermore, haemosiderin and ochronotic pigment in macrophages (fig 1F), and focal inflammatory infiltrates of lymphocytes and plasma cells with some lining layer hyperplasia and hypervascularity can be seen.1,2

E Kruithof, D Baeten, E M Veys, F De Keyser
Department of Rheumatology, Ghent University Hospital, Belgium

S Suykens, L De Wilde, R Verdonk
Department of Orthopaedics, Ghent University Hospital, Belgium

Correspondence to: Dr E Kruithof, Department of Rheumatology, OK12 IB, Ghent University Hospital, De Pintelaan 185, 9000 Gent, Belgium; elli.kruithof@uGent.be

References

Figure 1 (A) Darkening of the urine by standing; (B) x ray picture of the right shoulder demonstrating a destroyed joint with sclerosis, osteophytosis; (C) slate-blue discoloration of the antihelix of the ear; (D) yellow-brown shards, scattered over the synovium; (E, F) shard evoking a foreign body reaction with multinucleated giant cells and surrounded by pigment loaded macrophages.
Case Number 29: Ochronosis: synovial histopathological characteristics

E Kruithof, D Baeten, E M Veys, F De Keyser, S Suykens, L De Wilde and R Verdonk

*Ann Rheum Dis* 2004 63: 130
doi: 10.1136/ard.2003.013912

Updated information and services can be found at:
http://ard.bmj.com/content/63/2/130

These include:

References
This article cites 1 articles, 0 of which you can access for free at:
http://ard.bmj.com/content/63/2/130#BIBL

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/