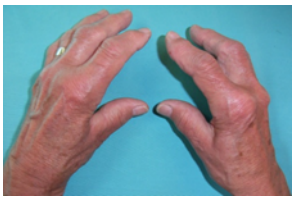
Could it be the arthritis that occurs in Haemochromatosis ?

Introduction:
Patients with Genetic Haemochromatosis (GH) may experience symptoms for up to 7 years or more before the diagnosis finally becomes apparent. This delays the commencement of treatment to reduce total body iron, usually by venesection, which is done to prevent hepatocellular carcinoma, hepatic cirrhosis, diabetes and cardiac failure.

Joint pain is very common, with over 75% of people reporting this pre-diagnosis. The following features are characteristic of arthritis in haemochromatosis (HA) and are sufficiently distinctive to enable earlier diagnosis of GH.

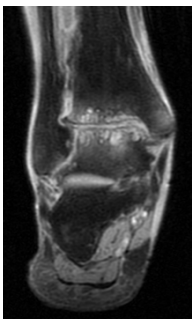
Demographics
Age of onset of joint pain is early, usually in the 5th decade, but can be as early as the 3rd decade.

Symptoms
Joint pain, sometimes with early morning stiffness, may be accompanied by unexplained fatigue.
An absence of trauma or other predisposing explanation is usual.



Clinical features
In HA, there is reduced range of movement and more bony rather than soft tissue swelling of affected joints. Involvement of joints in atypical sites for osteoarthritis (OA), such as the ankles and 2nd and 3rd metacarpophalangeal (MCP) joints may be seen. This can make it difficult to form a fist and sometimes results in the so-called 'iron fist'. HA can also involve typical sites for OA such as the proximal interphalangeal (PIP) joints, hips and knees.

Imaging features – the imaging signs mostly resemble those **of osteoarthritis**
X-ray: prominent osteophytes, described as 'hooks' at MCP joints, emerging from metacarpal heads, and at the 1st metatarsophalangeal joints emerging dorsally. Subchondral cysts are a frequent feature and chondrocalcinosis (mostly wrist and knee) is sometimes seen.
MRI: numerous and / or large bone marrow lesions (mainly cysts), full thickness cartilage loss and osteophytes.



Conclusion
Suspect iron overload in anyone below age 60 with presumed OA if you find any one or more of the following features:

- Fortuitous onset - no predisposing explanation, such as physical trauma, including ligamentous injuries, congenital abnormality, pre-existing inflammatory arthritis or family history.

- Finger MCP joint involvement, especially of the 2nd and 3rd MCP joints and / or ankle joints
- Florid osteophytes and cysts on imaging.
- Fatigue

If you identify a patient with these features, possibly accompanied by abnormal liver enzymes, check for iron overload by measuring transferrin saturation and ferritin, and / or refer the patient to a rheumatologist.

References

1. Richardson A, Prideaux A, Kiely PDW. Haemochromatosis: unexplained MCP or ankle arthropathy should prompt diagnostic tests; findings from two UK observational cohort studies. Scand J Rheumatol 2017; 46: 69-74. DOI: 10.3109/03009742.2016.1155645
2. Sahinbegovic E, Dallos T, Aigner E, Axmann R, Manger B, Englbrecht M et al. Musculoskeletal disease burden of hereditary haemochromatosis. Arthritis Rheum 2010; 62: 3792-8.
3. Carroll GJ, Breidahl WH, Bulsara MK, Olynyk JK. Hereditary haemochromatosis is characterised by a clinically definable arthropathy that correlates with iron load. Arthritis Rheum 2011; 63: 286-94

The HARI Group

Dr Axel Braner, Rheumatologist, University of Frankfurt, Frankfurt, Germany
Dr Barbara Butzeck, Patient Representative, HVD Germany, EFAPH France
Dr Graeme Carroll, Rheumatologist, University of Notre Dame, Freemantle, Australia
Dr Stephanie Finzel, Rheumatologist, University of Freiburg, Freiburg, Germany
Prof Pascal Guggenbuhl, Rheumatologist, Université Rennes, Rennes, France
Dr Helen Keen, Rheumatologist, University of Western Australia, Perth, Australia
Dr Patrick Kiely, Rheumatologist, St George's Healthcare NHS Foundation Trust, London, UK
Prof Gyorgy Nagy, Rheumatologist, Semmelweis – University Budapest, Hungary
Dr Jochen Zwerina, Rheumatologist, Hanusch Hospital, Vienna, Austria

HARI has also published a patient advice article on 'Treatment of Haemochromatosis Arthropathy' and 'Osteoporosis and Genetic Haemochromatosis'.

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