Musculoskeletal disorders and iron overload disease: comment on the American College of Rheumatology guidelines for the initial evaluation of the adult patient with acute musculoskeletal symptoms

To the Editor:
The recent clinical guidelines for the initial evaluation of the adult patient with acute musculoskeletal symptoms, proposed by the American College of Rheumatology (1), provide useful information and a good review for clinicians. However, there is one important omission in these guidelines. Nowhere in the guidelines is hemochromatosis mentioned. Such a prevalent and potentially life-threatening disease certainly deserves to be considered in the evaluation of patients with musculoskeletal disorders.

Hereditary hemochromatosis is now thought to be the most common genetic disorder in the white population (2). Approximately 1 in 250 persons is homozygous for this disorder and will develop the characteristic clinical manifestations such as diabetes, cardiomyopathy, liver disease, endocrine dysfunction, and, most notable for this discussion, arthropathy or other musculoskeletal disorders (2). Although hereditary iron overload disorders have traditionally been thought of as occurring exclusively in whites, recent research by Barton et al (3) indicates that approximately 1 in 67 African-Americans is affected by an etiologically distinct and severe form of iron overload. Hereditary iron overload disorders have been detected in persons of every ethnic background.

Arthropathy affects up to 80% of iron-overloaded patients and is often the only manifestation of this disease (4). Joint pain is a common and early symptom of iron overload, and “bone pain” has also been described as a common initial complaint (5). Clinically and radiographically, hemochromatotic arthropathy can resemble osteoarthritis, calcium pyrophosphate dihydrate deposition disease, pseudogout, rheumatoid arthritis, ankylosing spondylitis, or generalized osteopenia with osteoporoitic fractures (4,6,7). Since iron overload can cause such a wide array of musculoskeletal manifestations and because definitive clinical differentiation of iron overload from other arthropathies is very difficult, patients with peripheral arthropathy should be screened for iron overload. Indeed, recent research by Olynyk et al (8) indicates that the prevalence of iron overload is 5 times higher in patients with peripheral arthropathy than in the general population. Therefore, screening of patients with peripheral arthropathy for the possible presence of iron overload is justified.

Thus, since iron overload affects such a large portion of the population and arthropathy is a common manifestation of this disorder, patients with musculoskeletal symptoms should be screened for iron overload (4,8). The current literature suggests that everyone should be screened for iron overload even if there are no symptoms (8-10).

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