Case Study – Hereditary Hemochromatosis Associated Arthritis

Names and other identifying information have been changed to protect patient confidentiality.

Sarah, 53 year old female

Two and a half years ago, Sarah started having pain and swelling in her metacarpal joints and knees. She consulted several rheumatologists and received an extensive diagnostic workup without any specific cause being found. Her condition became progressively worse. She was treated with NSAIDS with mild improvement in her symptoms.

Sarah signed up for ActX testing because her family was signing up and not because of her arthritis. ActX testing showed that she had two C282Y variants (homozygous) in the gene HFE, variants associated with hereditary hemochromatosis.*

Further testing of Sarah showed high iron saturation and a moderately elevated ferritin, indicating some iron overload. The usual treatment for hereditary hemochromatosis is repeated phlebotomy, starting with ½ or 1 unit weekly, depending on the patient's weight, to reduce the patient's iron stores. Sarah was placed on weekly ½ unit phlebotomy.

Outcome

At the end of six weeks, her joint pain and swelling had resolved and Sarah was able to stop her medication.

(*Note:* Not all cases of arthritis associated with hereditary hemochromatosis respond well to phlebotomy, but in this case the response was good. Phlebotomy in patients with clear laboratory evidence of iron overload is considered effective in preventing a patient from developing outcomes such as hepatic cirrhosis and cardiomyopathy.)

Additional information on Hereditary Hemochromatosis

Hereditary hemochromatosis is an iron overload syndrome. The protein encoded by HFE is involved in the regulation of hepcidin synthesis. Hepcidin is the principal iron-regulatory hormone. It directs intestinal iron absorption and the retention of iron in macrophages. Variants in HFE lead to decreased levels of hepcidin, resulting in increased absorption of dietary iron, elevated serum iron levels, and iron retention in organs. Iron overload can lead to hepatic cirrhosis, cardiomyopathy, arthritis, progressive increase in skin pigmentation, diabetes mellitus, congestive heart failure and/or arrhythmias, hepatocellular carcinoma, and hypogonadism. Not all patients with C282Y variants develop iron overload, which is usually measured by ferritin levels and iron saturation.

References

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