

## QUESTION

A 65-year-old male with a long history of hypertension and hyperlipidemia presents with a 2-week history of increasing fatigue and weakness. He reports a recent weight loss of approximately 10 pounds (4.5 kg) and decreased appetite. He has no chest pain, shortness of breath, or palpitations. He has no history of smoking or alcohol use. He is currently on lisinopril and atorvastatin.

On physical examination, he appears thin and well-appearing. His vital signs are stable. There is no tachycardia or murmurs. The lungs are clear, and there is no lower-extremity edema. He has a normal neurological examination. Laboratory studies show a hemoglobin of 11.5 g/dL, hematocrit of 35%, and mean corpuscular volume of 100 fL. His serum ferritin is 100 ng/mL, and his serum iron is 150 µg/dL. His total iron-binding capacity is 300 µg/dL, and his transferrin saturation is 50%. His erythropoietin level is 10 U/mL. His renal function is normal, and his liver enzymes are within normal limits.

## ANSWER

The patient's presentation and laboratory findings are consistent with a diagnosis of iron overload. The key features include a high ferritin level (100 ng/mL), a high serum iron level (150 µg/dL), and a high transferrin saturation (50%). The patient's normal renal and liver function, as well as his normal erythropoietin level, further support this diagnosis. The patient's symptoms of fatigue and weakness, along with his weight loss, are likely due to the effects of iron overload on his organs.

The most likely cause of iron overload in this patient is hereditary hemochromatosis, a genetic disorder characterized by excessive iron absorption from the diet. This condition is caused by mutations in the HFE gene, which leads to increased iron absorption and accumulation in the body.

## DISCUSSION



The patient's symptoms and laboratory findings are consistent with a diagnosis of iron overload.