A 39 year old woman with chronic anemia and pain and swelling in her ankles. What is the diagnosis?
A 39-YEAR-OLD WOMAN WITH β-THALASSEMIA MAJOR HAD REQUIRED A transfusion of approximately 1 unit of red blood cells per month since she was 1 year old. Because of iron overload, chelation therapy was started when she was 6 years old. She presented with pain, swelling, and decreased joint mobility in both ankles. Laboratory evaluation was notable for a hemoglobin level of 8.3 g per deciliter, a mean corpuscular volume of 81.3 μm³, and a red-cell distribution width of 26.7%. Her older brother had also had β-thalassemia major and died of heart failure at 31 years of age. Anteroposterior (Panel A) and lateral (Panel B) views of the right tibia and fibula show the characteristic coarsened trabecular pattern, cortical thinning, and a widened medullary cavity. The radiograph indicates the degree of marrow hyperplasia, which produces characteristic osseous changes. This patient continued to receive transfusions with chelation therapy but died 1 year later because of heart failure.

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