The case:

A 20-year-old woman presented with foot pain after a minor injury.

Figure 1: Anteroposterior (A) and lateral (B) radiographs of the right foot and lateral radiograph of the spine (C).

Your diagnosis?

For answer see page 723
Diagnosis:
Osteopetrosis

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Answer to Radiologic Case Study
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A 20-year-old woman with a history of multiple fractures presented with foot pain after tripping in a pothole while walking. On physical examination, swelling and tenderness were observed in the dorsum of the foot. Anteroposterior and lateral radiographs of the foot showed increased sclerosis of all of the bones with a bone-in-bone appearance (Figure 2). A subtle, nondisplaced, incomplete fracture of the fifth metatarsal base was visible.

A radiograph of the spine obtained 5 years earlier for back pain shows diffusely increased sclerosis in the vertebral bodies with a sandwich vertebra appearance (Figure 3). The radiographic findings were consistent with the patient’s known diagnosis of osteopetrosis. The patient was treated with cast immobilization of the foot. At follow-up, the patient had no tenderness and the soft swelling had resolved. Repeat radiographs at that time demonstrated no evidence of a fracture, and the cast was removed.

BACKGROUND
Osteopetrosis is a rare sclerosing bone dysplasia and is the result of several rare genetic mutations. These mutations lead to varying degrees of dysfunction of the osteoclasts. This causes an imbalance between the osteoclasts and osteoblasts, resulting in excess bone formation and abnormal bone modeling. The end result is variable depending on the severity of osteoclast dysfunction and can range from asymptomatic adults with normal life expectancy to early infantile death.

Two genetic types of osteopetrosis are known: autosomal dominant and autosomal recessive. Autosomal dominant osteopetrosis, also known as osteopetrosis tarda or Albers-Schonberg disease, presents in adulthood. Most patients have a normal lifespan and are asymptomatic approximately 50% of the time. The current patient had a diagnosis of osteopetrosis tarda.

Autosomal recessive osteopetrosis, also known as osteopetrosis congenita, is seen in infancy to early childhood. It can cause bone marrow failure due to obliteration of the marrow cavity, as well as blindness and deafness due to narrowing of the bony neural

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IMAGING

Radiography and computed tomography are the most useful modalities to evaluate osteopetrosis. Magnetic resonance imaging can be helpful in more severe cases of autosomal recessive osteopetrosis to determine the amount of remaining marrow space. Bone marrow distribution of the disease is best shown by technetium-99m sulfur colloid scintigraphy.

On radiographs, osteopetrosis presents as dense bone or osteosclerosis. A characteristic radiographic appearance of osteopetrosis has been termed bone-in-bone, in which the dense central sclerosis appears as cortical bone inside the medullary cavity of the bone. This is most commonly seen in the iliac wing and epiphyses of the femur, humerus, tarsal bones (Figure 1), and ribs. In the vertebrae, dense sclerosis of the superior and inferior endplates can be observed and has been termed sandwich vertebra (Figure 2).

Autosomal recessive osteopetrosis presents early in infants. Pathological fractures are typical. The abnormality in bone modeling can result in undertubulation of the long bones, which can have a club-like shape or an Erlenmeyer flask appearance. Ribs tend to be sclerotic, and horizontal lucent lines may be present.

Individuals with autosomal dominant osteopetrosis tend to have symmetrical abnormalities. Two types, based on radiograph appearance, are observed. Type 1 is characterized by marked cranial sclerosis and calvarial thickening with minimal or no sclerosis in the vertebrae, patchy sclerosis in the pelvis, and diffuse sclerosis and cortical thickening in the tubular bones. Type 2 is characterized by a normal calvarium except for sclerosis at the skull base, vertebral involvement with thickening and sclerosis of the endplates, and dense sclerotic bones in the pelvis, especially along the iliac arcs. Transverse sclerotic banding is also common. Dental caries and abscesses, as well as sensory or motor loss in cranial nerves associated with foraminal hypertosis, are also indicators of type 2 autosomal recessive osteopetrosis.

TREATMENT

Treatment for osteopetrosis is largely based on symptoms. Autosomal dominant osteopetrosis requires no treatment unless pathologic fractures exist. However, more than half of all patients diagnosed with autosomal dominant osteopetrosis will need orthopedic surgery at some time. Because of the abnormal bone remodeling, delayed union or nonunion of fractures are common problems. During surgical fixation of fractures in these patients, problems can be encountered due to the hard and brittle bones, including perioperative fracture at the site of stress risers, such as screw holes and broken drill bits.

Due to the increased complications of surgery, casting of fractures is the preferred treatment when possible. Patients with autosomal recessive osteopetrosis may need red blood cell and platelet transfusions, and haematopoietic stem cell transplantation in severe cases of bone marrow dysfunction. Interferon-γ can improve immunity, increase bone resorption, and enlarge the marrow space. Other therapies that can be useful in stimulating osteoclast function in autosomal recessive osteopetrosis include calcium restriction and treatment with calcitriol, steroids, and parathyroid hormone.

REFERENCES