Case 2825
Sickle Cell Disease

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Section: Muskuloskeletal System
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Patient: 48 year(s), female

Clinical Summary

Female patient with past medical history of anaemia associated with multiple painful crises, characterized by severe skeletal pain and fever.

Clinical History and Imaging Procedures

Female black patient, 48 years-old, admitted in our hospital at age 8 with the diagnosis of Sickle Cell Disease. Her past medical history was remarkable for acute, painful vaso-oclusive crises (about 3 times a year) with fever; repeated episodes of pulmonary infections and tonsillitis; right hip arthroplasty for avascular necrosis of the femoral head at age 40; and gallstones, requiring a cholecystectomy at age 41. X-rays of the spine in both projections showed generalized decreased bone density, prominence of vertical trabeculae, and a stepped depression of the D9, D8 and D7 vertebral end plates (Fig. 1). It is also evident an oval mass compatible with calcified spleen, also seen at CT, and a coarsened trabecular pattern of the ribs. Frontal radiograph of the pelvis demonstrated osteoarthrosis of the right femoral head due to osteonecrosis (Fig. 2). CT scanning of the lumbar and sacral spine revealed distortion of the normal trabecular architecture, which results in a coarse trabeculation of the lumbar vertebras and sacrum. At the topogram it is possible to see the calcified spleen (Fig.3).

Discussion

Sickle Cell Disease (SCD) is an autosomal-recessive inherited disease in which defective sickle-shaped red cells are removed from the circulation and destroyed at increased rates, leading to anaemia. HbS, when deoxygenated, becomes relatively insoluble and forms aggregates with other haemoglobin

molecules within the RBC, causing vascular occlusion, which leads to tissue ischemia and infarction. SCD is a systemic illness with manifestations that range over nearly all organ systems. It is characterized early in life by severe chronic haemolytic anaemia caused by sickled haemoglobin and vaso-oclusion, bacterial infection, and organ infarctions, including the brain. The lung is a common site of involvement in SCD. Injuries range from acutes processes, such as pneumonia and Acute Chest Syndrome (ACS), to chronic entities, such as pulmonary fibrosis. ACS is defined by the occurrence of chest symptoms, new pulmonary infiltrate on chest radiograph, and in some cases fever, cough, dyspnoea, and chest pain. The radiographic finding necessary for the diagnosis of ACS is a single or multiple new areas of pulmonary consolidation (typically in the middle and lower lobe airspace). Repeated episodes of pulmonary insults can lead to chronic pulmonary disease (pulmonary fibrosis). Bone marrow necrosis, bone infarcts, osteomyelitis, and aseptic necrosis are common complications in patients with SCD. Expansion of the medullary space may be specially evident in the skull, which initially show a granular appearance followed by marked widening of the diploic space and thinning of the outer table (with sparing of the occipital), resulting a hair-on-end appearance. Hand-foot syndrome (dactylitis) is believed to be due to infarction of the red marrow and associated periosteal inflammation. Radiographs of the involved parts may show some periosteal elevation. Infarction (most common in long bones, but also seen in the spine, ribs, sternum, skull, clavicle, and facial bones) manifests over time, first with an ill-defined area of radiolucency, which subsequently develops arclike subchondral and intramedullary lucent areas, with patchy lucent and sclerotic areas. A peripheral rim of sclerosis is often seen, especially with medullary bone infarcts. Plain radiographs usually are not useful if obtained early in the course. In the spine, the pathognomonic ischemic vertebral findings are divided into 2 categories: the fish vertebra is a smooth biconcave deformity secondary to marrow hyperplasia with resultant osteoporosis, while the H vertebra is an abrupt cup-like lesion due to central infarction and collapse. The tower vertebra (an increase in vertical height of a vertebral body without an appreciable increase in girth) has been recently reported in some cases of SCD. At CT, an infarct initially manifests as disruption of the normal trabecular architecture and may be difficult to detect. As infarction progresses, it appears as circular areas of decreased attenuation. MR imaging is likely the most sensitive radiologic form of investigation, as it may show abnormality as soon as a few days after the ischemic insult Aseptic necrosis differs from bone infarction only in that is usual anatomic location, the articular surfaces and heads of long bones, can have consequences that are chronic, painful, and disabling. Approximately half of all patients with SCD will develop epiphyseal osteonecrosis by the age of 35 years. Initially, the patient has pain, but radiographs appear normal. Subsequent radiographs show some mottled attenuation of the epiphysis, subchondral lucent areas, and finally, flattening of the articular surface. Over time, osteoarthritis with joint space narrowing, articular surface sclerosis, and osteophytosis may occur. In weight-bearing joints, such as hip, joint replacement may be necessary. Osteomyelitis is a serious complication of SCD. It is most common in the diaphyseal region of bone and most often found in the femur, tibia and humerus. Although much less frequent than infarction, may be difficult to discriminate from infarction, even with clinical, laboratory, and radiological information. Radiographics findings of periostitis and osteopenia are non-specific and may be seen in both. The only definitive diagnostic test is a positive culture from blood

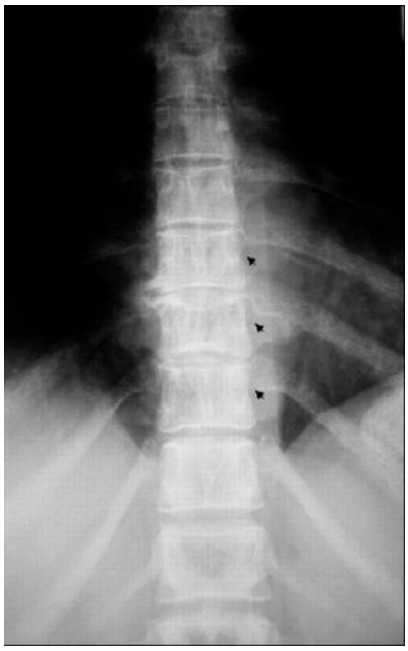
or bone puncture. The current major role of bone imaging is to guide placement of the needle so as to obtain a reliable specimen for culture. Other complications of SCD include leg ulcers, hepatobiliary manifestations (pigmented gallstones, cirrhosis, etc), functional autosplenectomy (which develops infection susceptibility), cardiac changes (electrical system fibrosis, ischemia), extramedullary haematopoiesis (although much more common in sickle cell variants; examples of sites which can occur: thorax, liver, spleen, skin, and adrenal glands), stroke, retinal ischemia, genitourinary problems (enuresis, chronic renal failure and priapism) Control of these complications is improving with advances in prophylactic antibiotic therapy, transfusion therapy, HbF induction, and bone marrow and stem cell transplantation.

Final Diagnosis

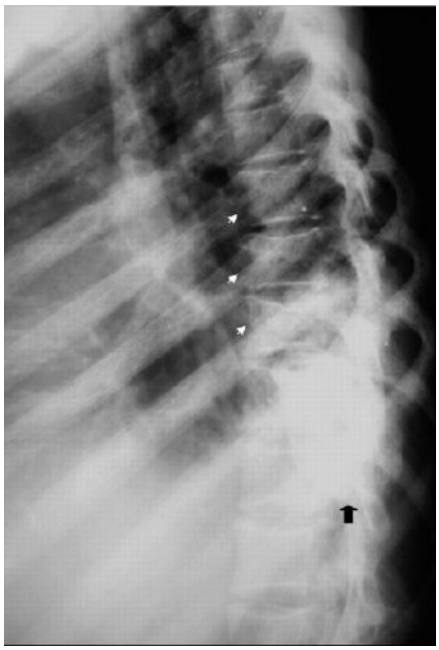
Sickle Cell Disease

Figures

Figure 1



Frontal X-ray of the dorsal spine showing generalized decreased bone density, prominence of vertical trabeculae, and a stepped depression of the D9, D8 and D7 vertebral end plates.

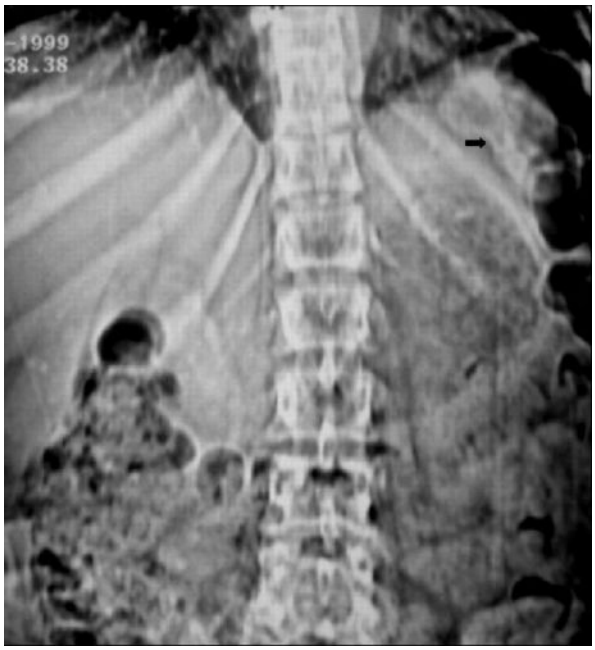


Lateral X-ray of the dorsal spine revealing generalized decreased bone density, prominence of vertical trabeculae, and a stepped depression of the D9, D8 and D7 vertebral end plates.

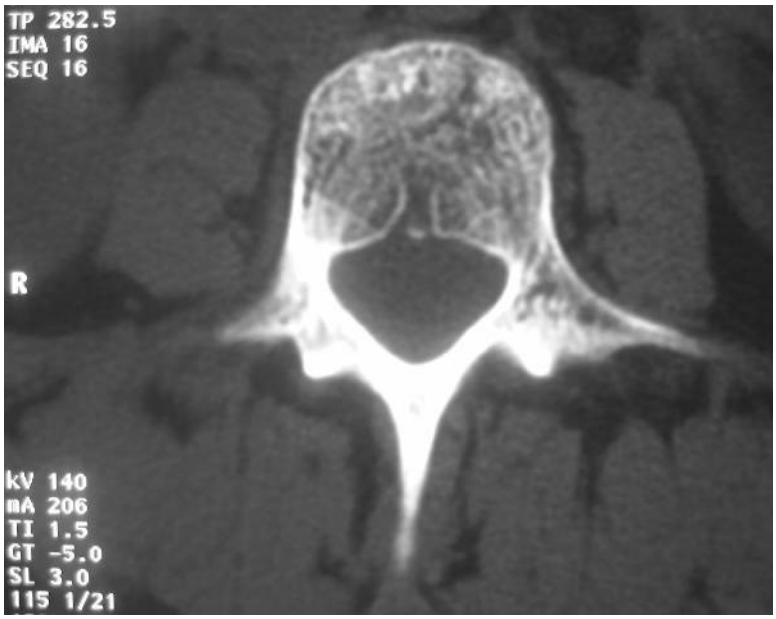
Figure 2

Osteoarthrosis of the right femoral head due to osteonecrosis.

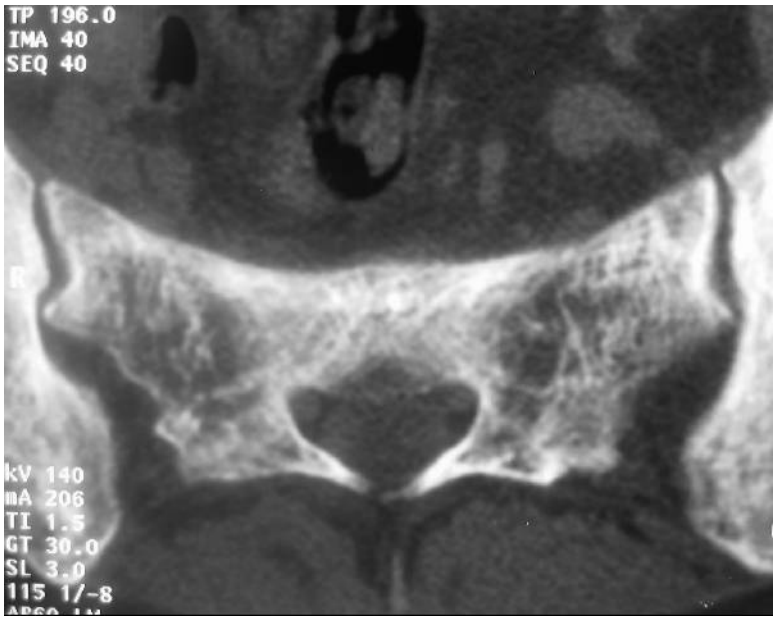
Figure 3



CT topogram of the abdomen showing a calcified spleen (black arrow).



CT scanning of the lumbar and sacral spine revealing distortion of the normal trabecular architecture, which results in a coarse trabeculation of the lumbar vertebra.



CT scanning of the lumbar and sacral spine showing distortion of the normal trabecular architecture, which results in a coarse trabeculation of the sacrum.

MeSH

Bone and Bones [A02.835.232]

A specialized CONNECTIVE TISSUE that is the main constituent of the SKELETON. The principle cellular component of bone is comprised of OSTEOBLASTS, OSTEOCYTES and OSTEOCLASTS, while FIBRILLAR COLLAGENS and hydoxyapatite crystals form the BONE MATRIX.

Anemia, Sickle Cell [C15.378.420.155]

A disease characterized by chronic hemolytic anemia, episodic painful crises, and pathologic involvement of many organs. It is the clinical expression of homozygosity for hemoglobin S.

References

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Citation

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