Outlines of Orthopaedic Pathology and Imaging

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A Review

By

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CHAPTER 1

METABOLIC BONE ABNORMALITIES

Osteoporosis

Definition:

- Generalized metabolic bone disease characterized by insufficient formation or increased resorption of bone matrix that results in a decreased bone mass and microarchitectural deterioration of bone, leading to an increased risk of bone fractures. Although there is a reduction in bone tissue, the tissue is fully mineralized (the bone is quantitatively deficient but qualitatively normal).

Epidemiology:

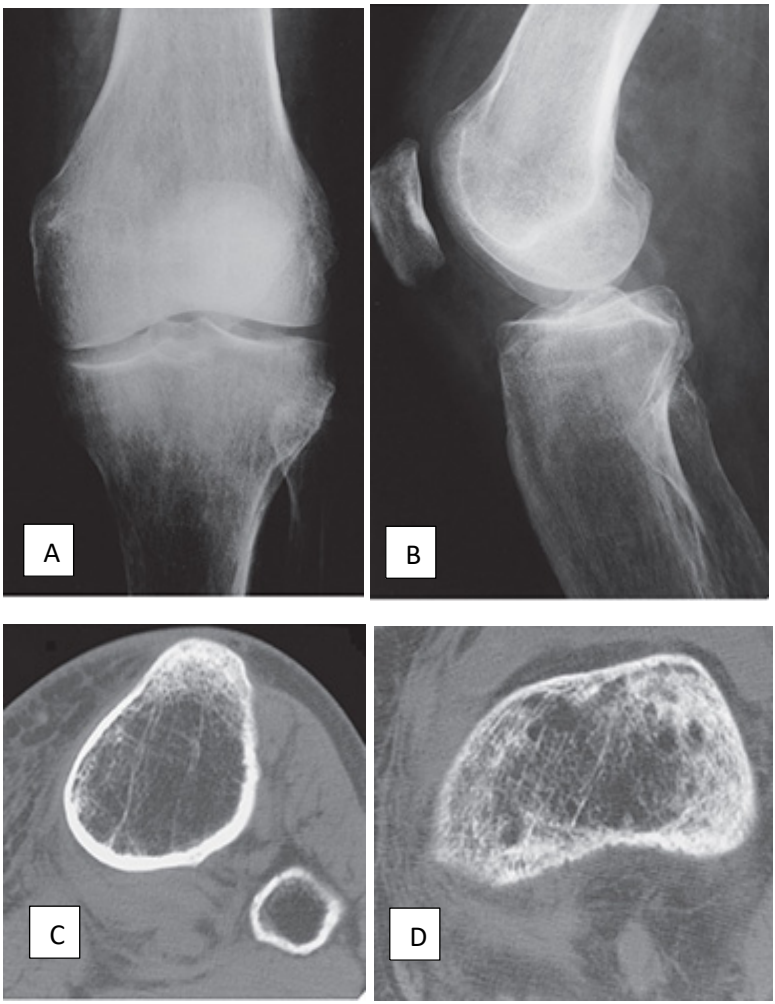
- Metabolic bone disease mostly affects patients older than 50 years.
- Commonly seen in postmenopausal women due to estrogen deficiency.
- Risk factors include poor nutrition (low calcium diet), aging, family history, medication, and immobilization.
- Major health problem affecting more than forty-four million Americans, with about two million fractures annually.

Clinical presentation:

- Most commonly affects the spine and pelvis.
- Vertebral fractures present with back pain, loss of height, and thoracic kyphosis.

Imaging:

- Diffuse osteopenia of bones (Figure 1).
- Thinning of the cortices of the long and short tubular bones.
- Dual-photon absorptiometry, Single X-ray absorptiometry, and Dual-energy X-ray absorptiometry (DEXA) are diagnostic.
- Occasionally, other methods are used, such as Digital computer-assisted X-ray radiogrammetry (DXR) or Quantitative ultrasound (QUS) techniques.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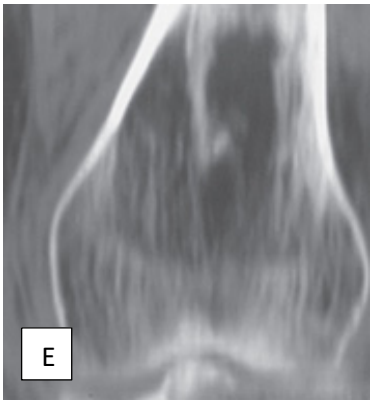

Figure 1. Anteroposterior (A) and lateral (B) radiographs of the knee **s**how increased radiolucency of bones, thinning of the cortices, and sparse trabecular pattern. These changes are more effectively demonstrated on axial computed tomography sections obtained through the proximal tibia and fibula (C) and distal femur (D), as well as on the reformatted coronal image of the distal femur (E). (Reprinted with permission of the Authors and Publisher from Greenspan A, Beltran J. Orthopaedic imaging – A practical approach. 7th ed., Philadelphia 2021, Wolters Kluwer, figure 27.2, p.1285).

Pathology:

Gross.

- Prominent loss of trabecular bone.
- Kyphosis of the thoracic spine.

Histopathology.

- Decrease of cortical and trabecular bone (Figure 2).
- Trabecular bone is more affected by prominent trabecular thinning, discontinuation, and separation (Figures 3 and 4).
- Bone cortex is thinning with widened Haversian channels.

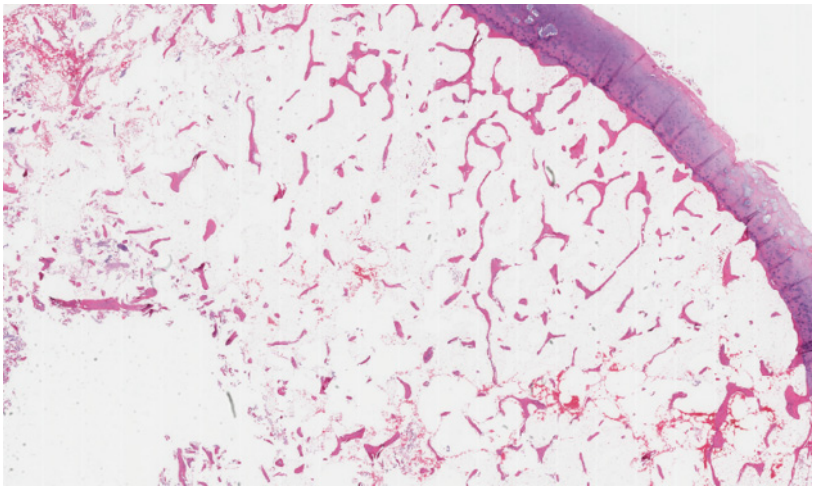

Figure 2. Decrease in cortical and trabecular bone.

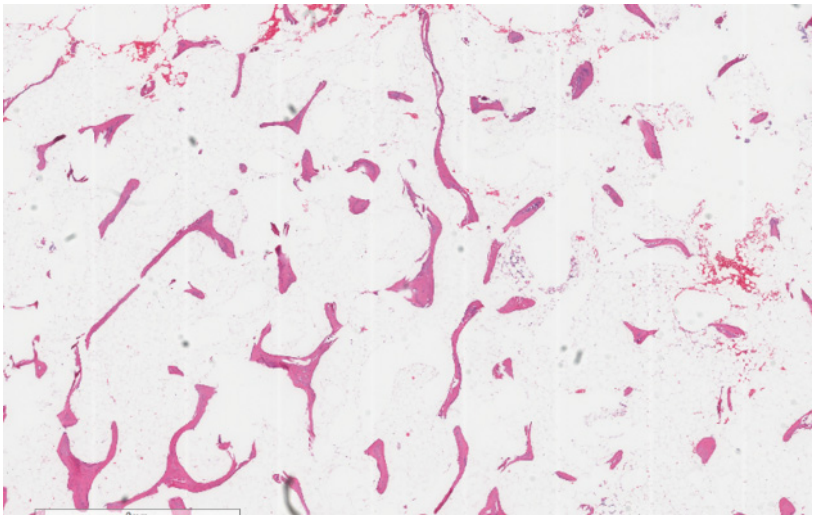

Figure 3. Trabecular bone thinning, discontinuation, and separation.

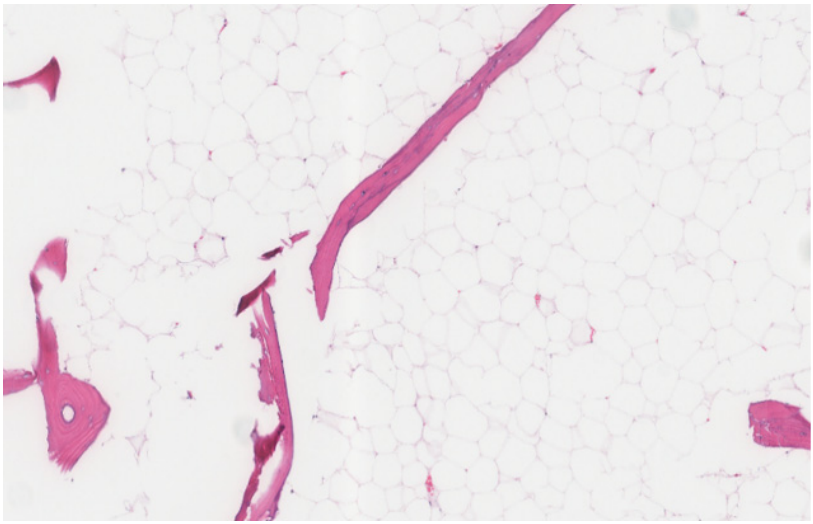

Figure 4. Thinning of bone trabeculae.

Prognosis and therapy:

- Pathologic fractures correlate with increased mortality.
- The therapeutic goal is to prevent bone loss and fractures with proper nutrition (vitamin D and calcium supplementation), exercise, and medication (bisphosphonates, estrogen receptor modulators such as raloxifene).

Differential diagnosis:

- Osteomalacia.
- Osteogenesis imperfecta.
- Hyperparathyroidism.

Osteomalacia

Definition:

- Defective skeletal mineralization (calcification) of a bone matrix with an accumulation of unmineralized bone caused most often by faulty absorption of fat-soluble vitamin D from the gastrointestinal tract secondary to malabsorption syndrome.

Epidemiology:

- Associated with bone and soft tissue tumors such as phosphaturic mesenchymal tumors.
- Deficiencies or disorders of phosphorus, calcium, and vitamin D metabolism.

Clinical presentation:

- Musculoskeletal weakness and bone pain are common features.
- Bone fractures of vertebral bodies and femoral neck are common findings.
- Low levels of vitamin D and increased alkaline phosphatase.
- Milkman syndrome, a condition with numerous pseudo fractures.
- Association with neurofibromatosis, fibrous dysplasia, and Wilson disease.
- Oncogenic osteomalacia (also known as tumor-induced osteomalacia – TIO) is a paraneoplastic syndrome characterized by hypophosphatemia, hyperphosphaturia, and low plasma levels 1,25-dihydroxy vitamin D, caused by bone and soft-tissue tumors or tumor-like lesions.

Imaging:

- Multiple, bilateral, and often symmetrical radiolucent lines in the cortex, perpendicular to the long axis of the bone, referred to as *pseudo fractures* or *Looser zones*.
- Generalized osteopenia.
- Total decrease in bone mineral density on DEXA studies.
- Areas of unmineralized bones caused by fast bone resorption and slow mineralization.

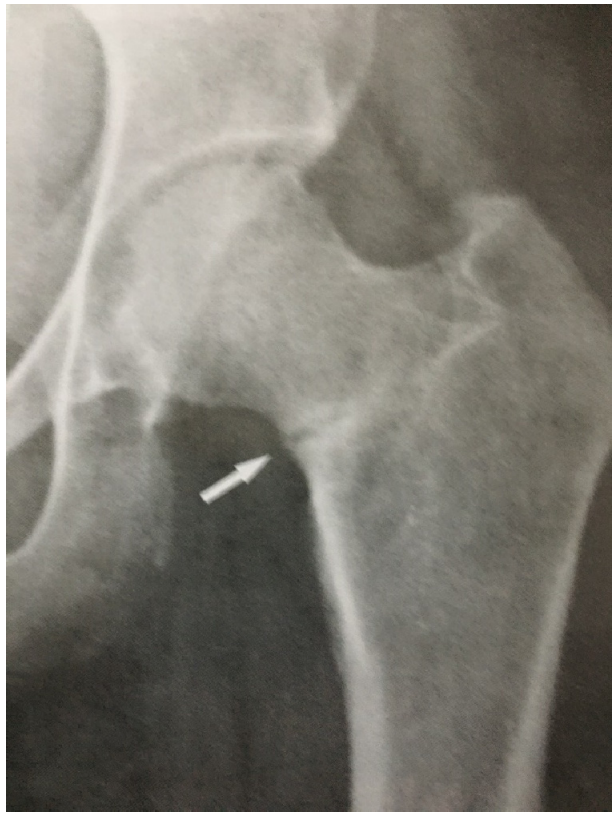

Figure 1. A looser zone, also called insufficiency-type stress fracture, represented by a radiolucent defect in the cortical bone, reflects the accumulation of non-mineralized osteoid tissue and is a characteristic feature of osteomalacia. (Reprinted with permission of the Authors and Publishers from Greenspan A, Beltran J. Orthopedic imaging. A practical approach. 7th Ed. Philadelphia, 2021, Wolters Kluwer, Fig. 26.5, p. 1276).

Pathology:

Gross.

- Soft unmineralized bone, easy to fracture.

Histopathology.

- Excessive quantities of inadequate mineralized bone matrix (osteoid) coating the surface of trabeculae in spongy bone and lining the Haversian canals in the cortex.
- Decreased mineralization and calcification of the bone.
- Von Kossa stain highlights an unmineralized portion of the bone trabeculae (eosinophilic) and a mineralized portion (dark grey or black) (Figure 2).

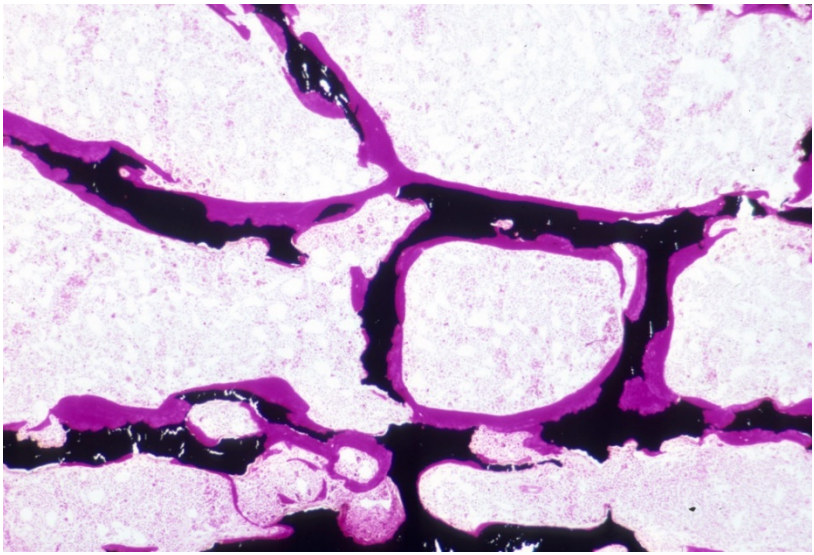

Figure 2. Von Kossa stain highlights an eosinophilic unmineralized portion of the bone at the surface of the bone. (Courtesy of Michael Klein, M.D., New York).

Prognosis and therapy:

- Main cause of bone fractures in elderly patients.
- Correction of vitamin D deficiency or underlying medical condition as treatment.

Differential diagnosis:

- Osteopenia.
- Osteoporosis.
- Hyperparathyroidism.

Hyperparathyroidism

Definition:

- *Hyperparathyroidism* is a disease occurring because of overactivity of parathormone-producing parathyroid glands resulting in excessive secretion of parathyroid hormone – PTH (which regulates calcium metabolism and has an impact on osteoclasts and osteoblasts activity) either secondary to gland adenoma (90% of cases), gland hyperplasia (9% of cases) or carcinoma (1% of cases). In addition, increased secretion of PTH causes secondary hyperparathyroidism in response to a sustained hypocalcemic state associated with renal disease or malnutrition (severe calcium or vitamin D deficiency). Brown tumors commonly occur in primary and secondary hyperparathyroidism.

Epidemiology:

- Mostly seen in 50-year-old patients and menopausal women.
- In patients with multiple endocrine neoplasias, type I, and type IIA.

Clinical presentation:

- Bone and joint pain with pathologic fractures due to osteopenia.
- Weakness, muscular hypotonia, nausea, anorexia, constipation, polyuria, and thirst.
- Nephrocalcinosis.
- Depression and forgetfulness.
- Laboratory studies show increased levels of calcium and decreased levels of phosphorus.
- Long-standing form results in forming multiple large cystic lesions in bones (osteitis fibrosa cystica).

Imaging:

- General osteopenia.
- In the skull, the characteristic mottling of the vault yields a "salt-and-pepper" appearance.
- Subperiosteal, subchondral, and intracortical (tunneling) bone resorption (Figure 1).
- Resorption of acromial ends of the clavicles.
- Loss of lamina dura around the tooth socket.
- Brown tumors (also known as osteoclastomas) present circumscribed lytic lesions resulting from massive bone resorption due to osteoclast activity, associated with hemorrhage and hemosiderin deposition (brown color). They may be solitary or multifocal, resembling metastases. Commonly they involve facial bones, ribs, pelvic bones, and long and short tubular bones (Figures 2 and 3). They can occur in primary and secondary hyperparathyroidism and renal osteodystrophy.
- In secondary hyperparathyroidism, soft tissue and cartilage calcifications, bone density increase, and the spine have a characteristic sandwich-like appearance (rugger-jersey spine).

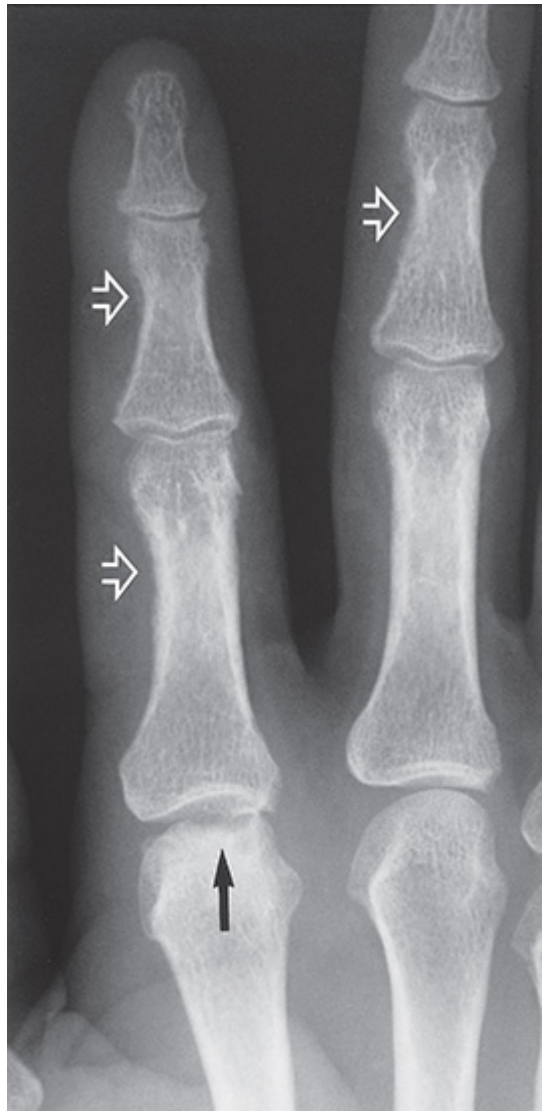

Figure 1. The radiograph of the finger shows subchondral resorption at the head of the second metacarpal bone (arrow) and subperiosteal resorption at the proximal and distal phalanges (open arrows).

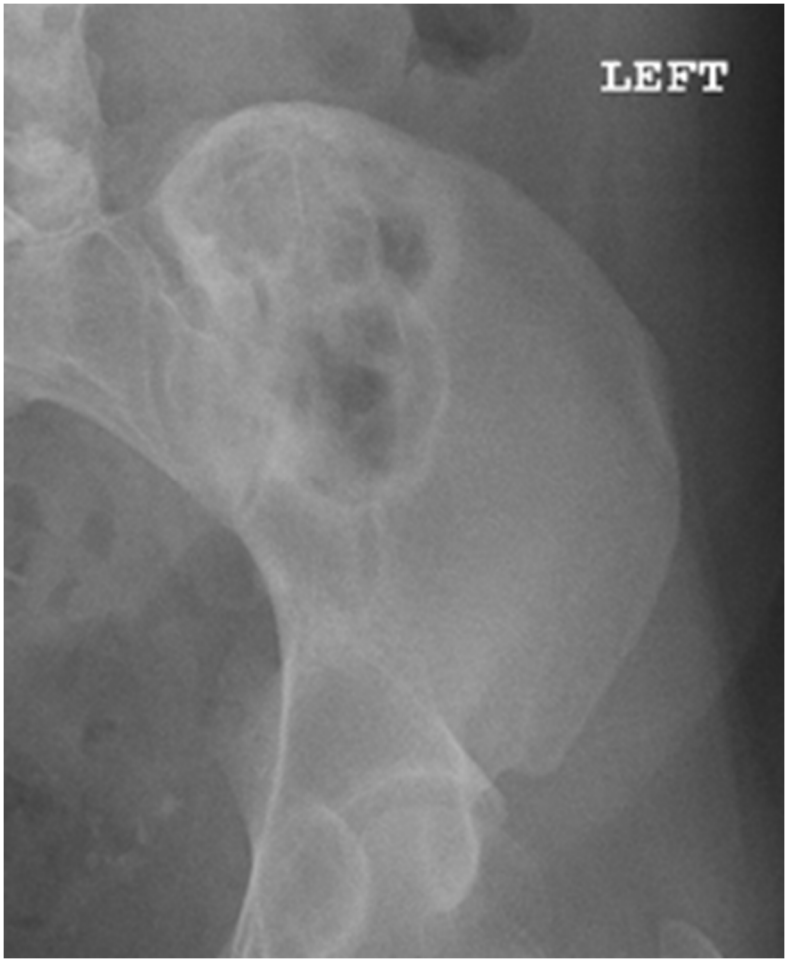

Figure 2. A radiograph of the ilium shows well-circumscribed lytic lesions representing brown tumors.

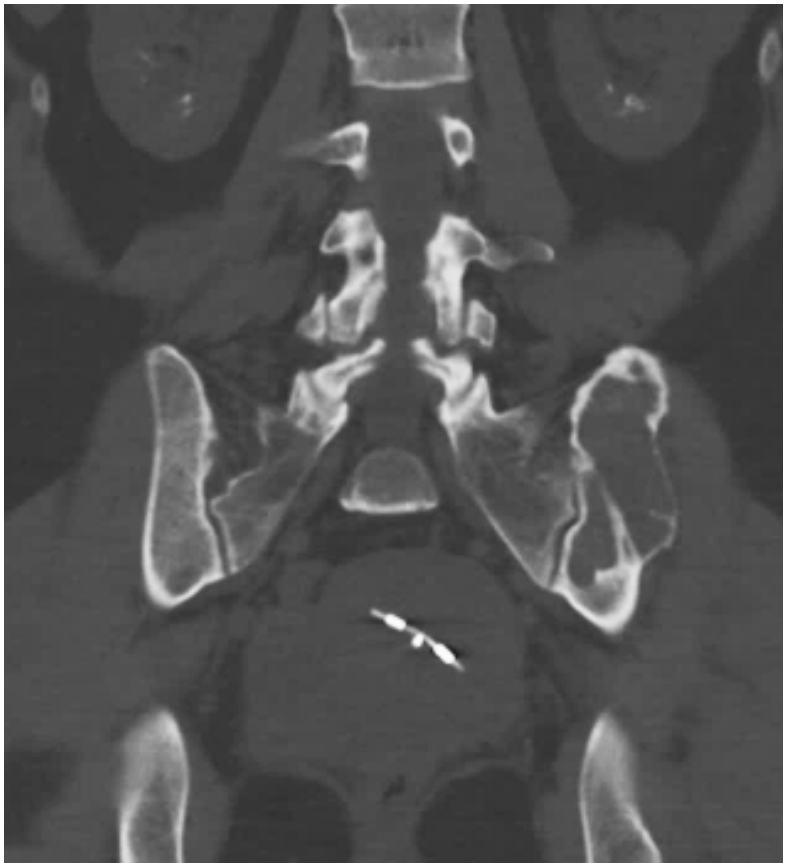

Figure 3. A coronal reformatted CT image of the pelvis shows a sizeable low-attenuation lesion within the left ilium representing a brown tumor.

Pathology:

Gross.

- Fragments of friable brown tissue.

Histopathology.

- Uniform spindle histiocytic cell proliferation with aggregates of osteoclast-like giant cells and hemosiderin deposition (Figure 4).
- Increased osteoclastic activity with intertrabecular tunneling (dissecting resorption) and bone resorption (Figure 5).
- Abundant fibrosis, hyalinization, and bone sclerosis can be found (Figure 6).
- Histopathology of the brown tumor may resemble giant cell reparative granuloma.

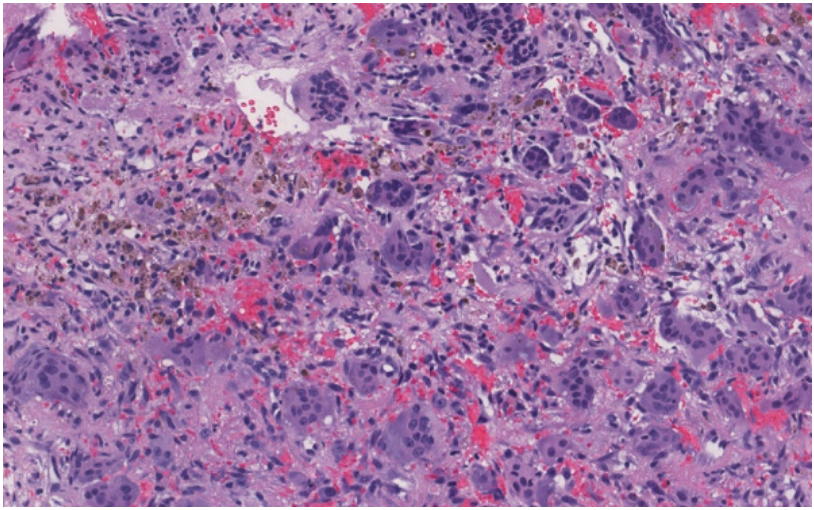

Figure 4. Uniform spindle histiocytic cell proliferation with aggregates of osteoclast-like giant cells and hemosiderin deposition.

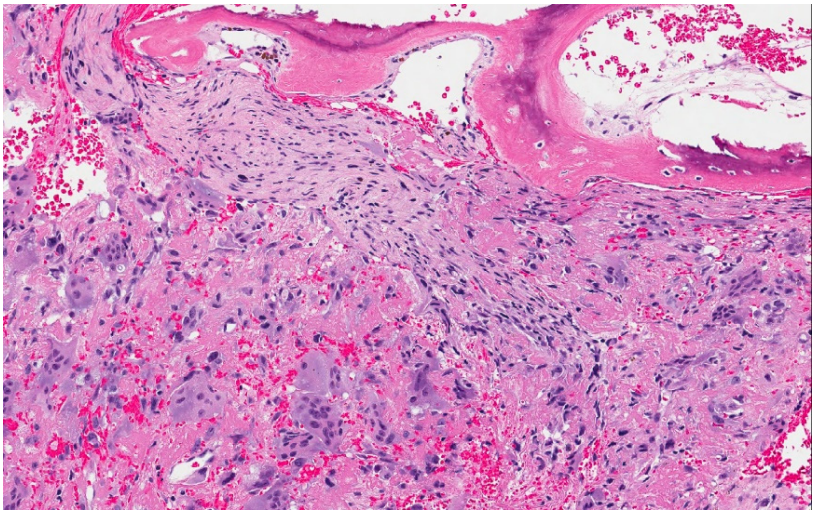

Figure 5. Increased osteoclastic activity with intertrabecular tunneling and bone resorption.

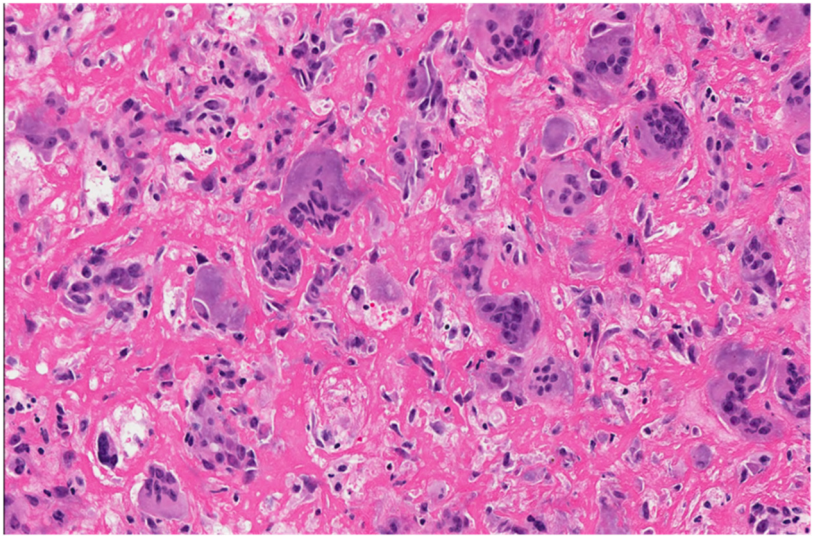

Figure 6. Stromal fibrosis and hyalinization.

Prognosis:

- Surgical removal of the hyperfunctioning parathyroid gland is curative.

Differential diagnosis:

- Myelofibrosis.
- Giant cell tumor of bone.
- Giant cell reparative granuloma.
- Lytic metastases.
- Osteomalacia.

Alkaptonuria - ochronosis

Definition:

- A rare autosomal recessive inherited disease characterized by the presence of homogentisic acid in the urine, which turns black when oxidized (alkaptonuria), and deposition of dark pigment in cartilage or fibroconnective synovial tissue (ochronosis) caused by lack of the enzyme homogenous acid oxidase which plays a part in the normal degradation process of the aromatic amino acids tyrosine and phenylalanine.

Epidemiology:

- Affects males and females equally.
- More common in certain areas in Slovakia, Dominican Republic, Jordan, and India.
- Most affected are large joints, including knees, shoulders, and hips.

Clinical presentation:

- In general, affected individuals are asymptomatic until adult life.
- Mild pain and decreased range of motion in the affected joints.
- Osteoarthritis of large joints and spine.
- Gray to brown pigmentation in the sclera of eyes.
- Thickened and darkened cartilage in the ears.
- Discoloration of the skin, particularly around sweat glands.
- Dark-colored urine and kidney stones.

Imaging:

- Dystrophic calcifications in the intervertebral disk spaces, articular cartilage, tendons, and ligaments.
- Degenerative changes of the hip, spine, and knee joints with joint and disk space narrowing.
- Calcification and fusion of disk spaces (Figure 1).
- Imaging appearance may mimic osteoarthritis and CPPD crystal deposition arthropathy.

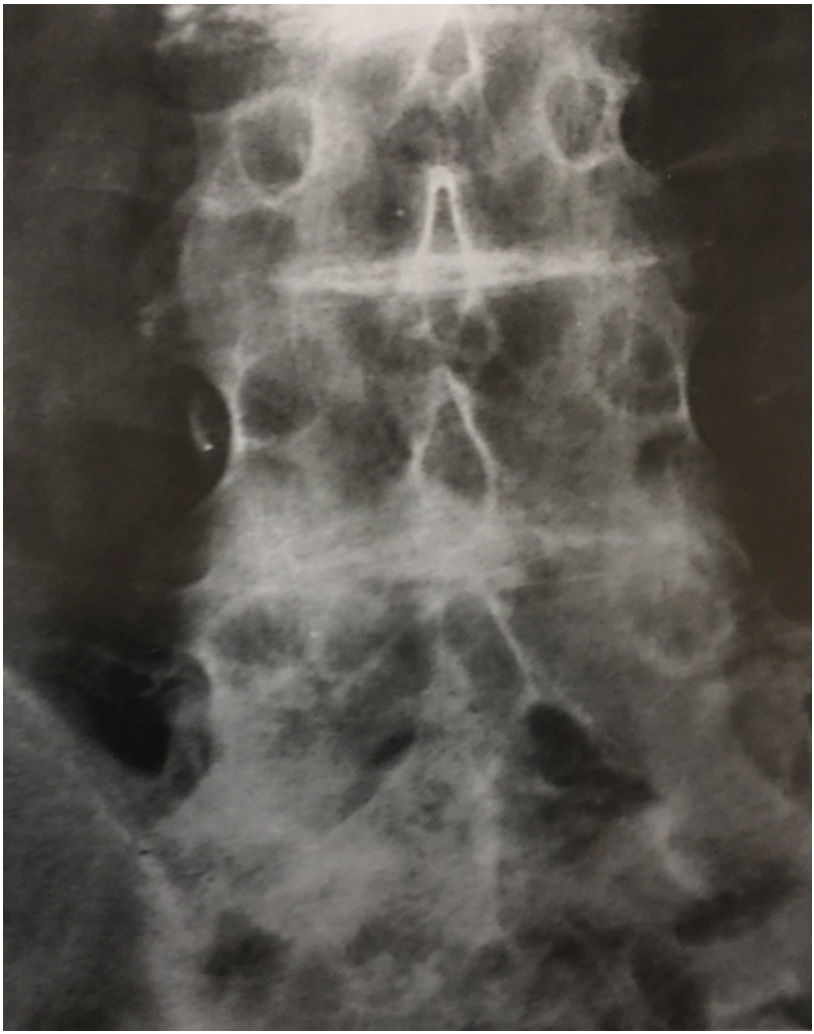

Figure 1. A lumbar spine radiograph demonstrates the narrowing of several intervertebral disk spaces associated with characteristic calcifications. (Reprinted with permission of the Author and Publisher from Greenspan A, Gershwin ME. Imaging in rheumatology. A clinical approach. 1st ed. Philadelphia, 2018, Wolters Kluwer, Fig. 7.42, p.298).

Pathology:

Gross.

- Black pigmentation of skin, cartilage, tendons, and ligaments.
- Cartilage is brittle and fragmented.

Figure 2. The sagittally sectioned spine specimen shows black pigmentation within the narrowed intervertebral disks.
(Reprinted with permission of the Author and Publisher from Greenspan A, Gershwin ME. Imaging in rheumatology. A clinical approach. 1st ed. Philadelphia, 2018, Wolters Kluwer, Fig. 7.41, p.298).

Histopathology.

- Dark pigmentation of cartilage and fibroconnective synovial tissue.

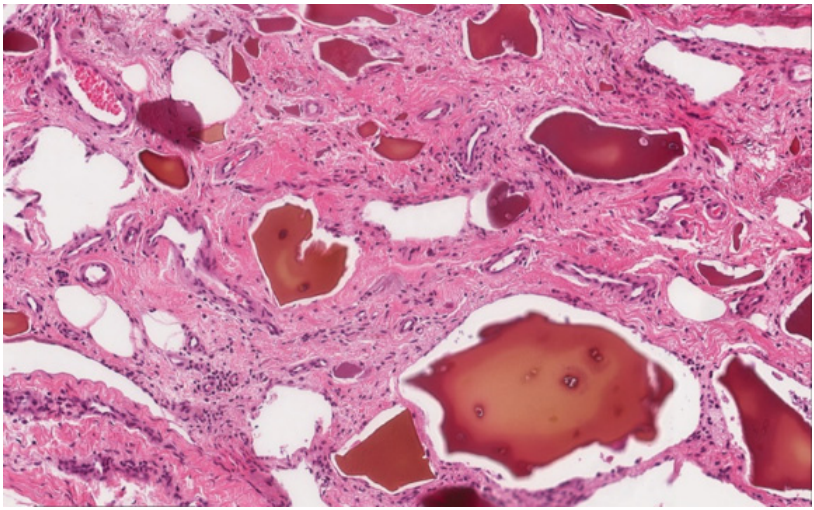

Figure 3. Dark pigmentation of cartilage and synovial tissue.

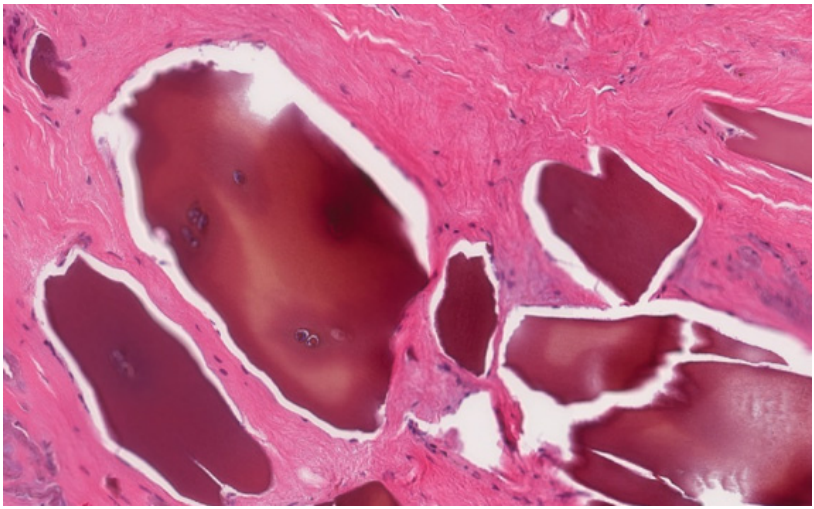

Figure 4. Dark pigmentation of the cartilage.

Genetics:

- The disease is caused by a mutation in the HGO gene located in the arm of chromosome 3q1, responsible for the enzyme homogentisate 1,2-dioxygenase, which leads to the accumulation of homogentisic acid (dark pigment) in various organs, with a predilection for connective tissue.

Differential diagnosis:

- Medication-induced hyperpigmentation (e.g., minocycline).
- Osteoarthritis.
- CPPD crystal deposition arthropathy.

Paget disease

Definition:

- Chronic, progressive disturbance in bone metabolism affects normal bone remodeling due to an imbalance between bone formation by osteoblasts and bone resorption by osteoclasts, leading to bone enlargement of bones and weakness followed by stress and frank fractures.

Epidemiology:

- Most patients are older than 55 years old, rare in patients younger than 40.
- Familial occurrences were reported.
- More common occurrence was reported in North America, Australia, New Zealand, Europe, and people of Anglo-Saxon descent.

Clinical presentation:

- Commonly involves the pelvis, spine, skull, and lower extremities.
- Frequently asymptomatic, particularly in early stages.
- Periosteal tenderness, bone pain, and arthritis.
- Neural compression.
- Long-standing disease can cause an increase in head size (headaches and deafness), bowing deformities of the limbs, and curvature of the spine (spinal stenosis).
- Elevated serum alkaline phosphatase levels with normal serum calcium and phosphorus levels.