

Albers-Schönberg disease

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A 39-year-old woman with no known systemic disease presented to the respiratory clinic for a routine chest radiography examination. She had no respiratory symptoms such as cough, sputum, dyspnea, or chest pain. Physical examination revealed bilaterally symmetrical lung sounds without crackles or wheezes. Chest radiography revealed diffuse osteosclerosis in the thoracic cage, bilateral clavicles, and scapulae, and cortical thickening of bilateral ribs (Fig. 1, arrows). Computed tomography of the chest at the mediastinum window (level 40 and width 350 Hounsfield units) revealed diffusely homogenous marked hyperdensities of all bony structures (Fig. 2). Dual-energy X-ray absorptiometry revealed a T-score of 5.6 (normal range, -1 to 1), obtained from L2 to L4 spines. Detailed anamnesis disclosed that her father had had a history of bilateral hip frac-

tures and been told that he had dense bones on radiographic examination. A diagnosis of Albers-Schönberg disease or autosomal dominant osteopetrosis was made. She was advised to avoid strenuous exercise for prevention of

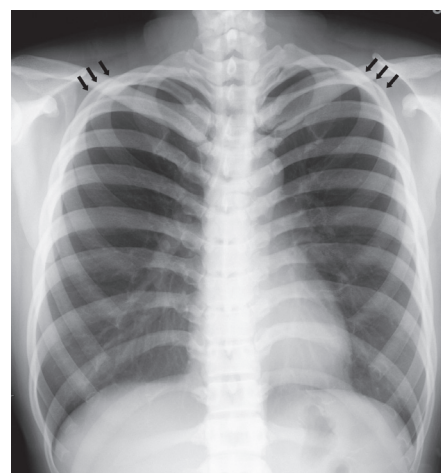


Figure 1. Chest radiography showing diffuse osteosclerosis in the thoracic cage, bilateral clavicles, and scapulae, and cortical thickening of bilateral ribs (arrows).

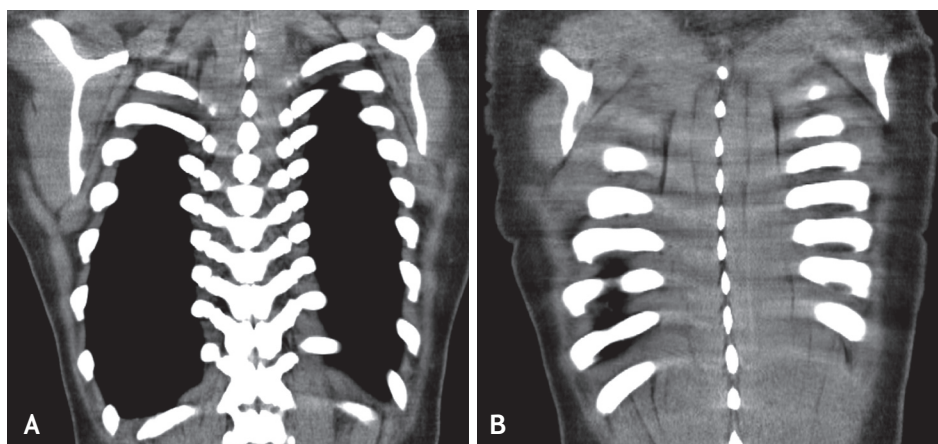


Figure 2. Computed tomography of the chest at the mediastinum window (level 40 and width 350 Hounsfield units) revealing diffusely homogenous marked hyperdensities of all bony structures. (A) Mid-coronal plane. (B) Posterior coronal plane.

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fracture and she remained asymptomatic with regular follow-up at the clinic.

Osteopetrosis is a heterogeneous hereditary disorder with a defect in bone resorption by osteoclasts and can be categorized into three primary types, including infantile osteopetrosis, intermediate autosomal recessive osteopetrosis, and autosomal dominant osteopetrosis, also known as Albers-Schönberg disease. In Albers-Schönberg disease, the complications are often confined to the skeleton, such as fractures, scoliosis, osteoarthritis, and osteomyelitis. The life expectancy is normal, and treatment is mainly supportive. The major differential diagnoses of generalized osteosclerosis

on chest radiography are osteoblastic metastases from prostate or breast cancer. The correct diagnosis could be obtained by detailed history taking that discloses the compatible family history for osteopetrosis and the absence of these cancers. Genetic investigation can be used to confirm the definite diagnosis, differentiate subtypes of osteopetrosis, and provide further prognostication.

The informed consent is confirmed.

Conflict of interest

No potential conflict of interest relevant to this article was reported.