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LETTER TO THE EDITOR

Hajdu–Cheney syndrome with ventricular septal defect

To the Editor:

Hajdu-Cheney syndrome (HCS) is a rare osteolysis syndrome characterized by generalized, progressive osteoporosis and acroosteolysis [1]. Cardiovascular abnormalities, including patent ductus arteriosus or atrial or ventricular septal defect (VSD) might be rarely accompanied by HCS as case presentations [2].

We present here a case of HCS with cardiovascular defect that has not been reported previously in the literature.

A 31-year-old man presented to our clinic with a non-healing wound on the plantar surface of his left foot. He had a history of right below-knee amputation, surgical closure of VSD, and spontaneous autoamputation of the right metatarsophalangeal joint and the first, third, and fourth distal phalanges of the left foot. His general condition was stable with blood pressure of 120/80 mmHg, pulse rate of 76/min, and his fever was 37.3°C. He had dysmorphic facial features with low-set ears, micrognathia, and diastema. He had two ulcerated and purulent wounds on the metatarsophalangeal joint of the plantar surface of the left foot. The sizes of the wounds were 2 × 2 cm and 7 × 1.5 cm that extended to all skin layers and bone (Fig. 1A). His laboratory test results were as follows: hemoglobin 11.4 g/dL; neutrophils 5.8 × $10^3/\mu L$; thrombocytes 433,000/μL; erythrocyte sedimentation rate 21 mm/h. Liver and kidney function tests were normal, and urinalysis, antinuclear antibody, rheumatoid factor, complement, and serum immunoglobulin levels were within normal range. Electrocardiography revealed right bundle-branch block. Echocardiography, chest radiography, and arterial and venous Doppler ultrasonography of the left lower extremity were normal. The proximal phalanges were deformed and amorphous, and no distal/medial phalanges

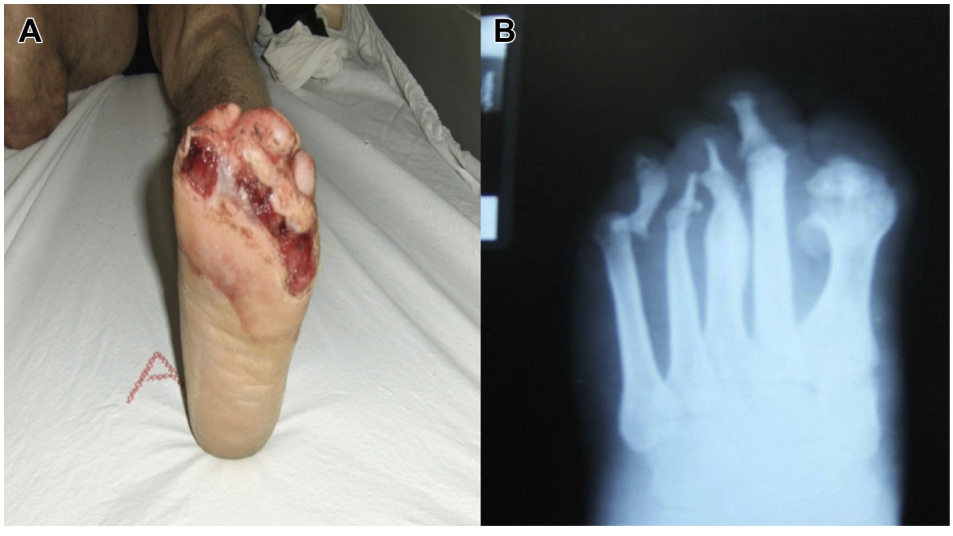

Figure 1. (A) Ulcerated and purulent wounds on the metatarsophalangeal joint of the plantar surface of the patient's left foot that are 2 × 2 cm and 7 × 1.5 cm that extended into all skin layers and bone. (B) Radiograph showing acroosteolysis.

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were seen on X-ray (Fig. 1B). Bone mineral density measurement showed signs of osteoporosis (total T-score of the lumbar vertebrae was −4.84). Biopsy of the wound revealed ulcer and granulation tissue on the ulcerous base.

Generalized osteoporosis is a typical symptom of this syndrome and is associated with rapid bone turnover. The genetic defect and molecular pathogenesis of HCS is not completely known. It may not consist of venous changes only. However, osteoblast dysfunction may explain the pathogenesis of the disease [1,3]. The molecular pathogenesis of the cardiovascular abnormalities of the syndrome is unknown [2]. Myxomatous degeneration or focal calcifications of mitral valve leaflet were reported. Kaler et al. [2] presented a case of HCS with severe mitral regurgitation and mild aortic stenosis. However, our survey of the literature revealed no cases of HCS with VSD, among almost 60 cases currently presented.

As a result, a careful anamnesis and clinical suspicion of this syndrome are the most important factors in an early diagnosis. Early diagnosis and treatment may improve prognosis for the patients. Therefore, acroosteolysis syndromes and also coexistence of VSD should be kept in mind for patients who presented at an early age and with progressive osteoporosis.

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