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Pachydermoperiostosis Accompanied by Heart Failure

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Pachydermoperiostosis or primary hypertrophic osteoarthropathy is an uncommon disease of acromegaloid facial feature, but characterized by unique phenotype (digital clubbing and pachydermia) and distinctive radiographic appearances like periostosis. We experienced a case with complete form of pachydermoperiostosis accompanied by heart failure. He presented with typical features consisting of clubbing with enlargement of the hand, thickening of facial skin and periosteal new bone formation involving lower leg. Echocardiography revealed severely decreased left ventricular systolic function. Treatment with medications resulted in an improvement of cardiac function and symptom. There is no previous report documenting pachydermoperiostosis accompanied by heart failure. We report that case for the first time.

KEY WORDS: Heart failure · Systolic · Echocardiography · Primary hypertrophic osteoarthropathy.

Introduction

Hypertrophic osteoarthropathy is characterized by the coexistence of digital clubbing and periosteal proliferation of the tubular bones. Pachydermoperiostosis or primary hypertrophic osteoarthropathy is clinically similar to acromegaly and is manifested by finger clubbing, hypertrophic skin changes and periosteal bone formation. Pachydermoperiostosis is a rare genodermatosis and occurs predominantly in men, who usually show a more severe phenotype. Three forms of pachydermoperiostosis are described: complete, incomplete and fruste form. The major diagnostic criteria include digital clubbing, periostosis and pachydermia.[1)]

There is no previous report documenting pachydermoperiostosis accompanied by heart failure. Here we report the case of the complete form of pachydermoperiostosis, who accompanied by heart failure.

Case

A 34-year-old male presented with complaints of exertional dyspnea since 5 days ago. He presented with 3 years history of hypertension. There was not any specific past medical history. On arrival in the emergency department, he had a pulse rate of 100 beats per minutes, blood pressure of 150/100 mmHg and respiration rate of 22 breaths per minutes. His electrocardiogram on admission showed left ventricular hypertrophy and normal sinus rhythm. A chest X-ray showed an increased cardiothoracic ratio in association with mild pulmonary congestion. Cardiac enzyme were normal, N-terminal pro B-type natriuretic peptide was increasing with 1143 pg/mL.

At initial physical examination, his acromegalic-look make to evaluate endocrine study. Results of laboratory analyses, including growth hormone, insulin-like growth factor 1, 75 g oral glucose tolerence test, thyroid-stimulating hormone, free-triiodiothyronine, free-thyroxine, were normal. His facial skin was greasy and thickening (deep frontal folds and heavy eyelids) (Fig. 1). His both hands had broad hands, clubbing of fingers, swollen interphalangeal joints and round

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turtle-back-shaped nails (Fig. 2). X-ray of bones showed periosteal new bone formation in the lower end of tibia, talus and calcaneus (Fig. 3). This physical exam and radiologic finding demonstrated specific features of pachydermoperiostosis.

Transthoracic echocardiography (TTE) revealed enlarged left ventricle (LV) (LV end-diastolic dimension=65.7 mm) and left atrium (LA) chamber dimensions, and decreased LV systolic function with severe global hypokinesia (LV ejection fraction (EF)=34.4%, end-diastolic volume/end-systolic volume=122.1 mL/83.1 mL) (Fig. 4.). Also TTE showed eccentric LV hypertrophy (213.3 g/m^2).

Treatment with diuretics and angiotensin converting enzyme inhibitor (ACE-I) resulted in an improvement of pulmo-

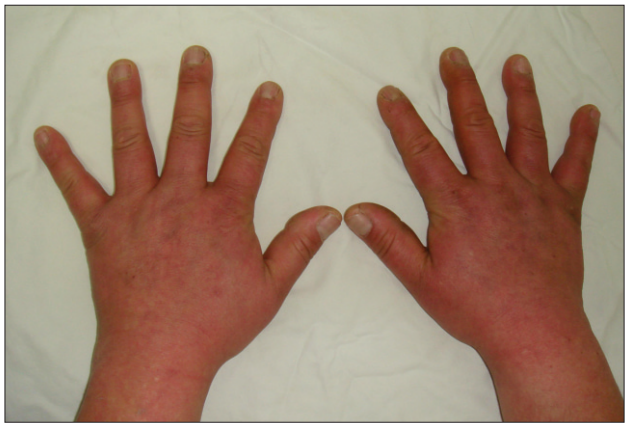

Fig. 2. Photograph of the patient's both hands reveals clubbing of fingers, swollen interphalangeal joints and round turtle-back-shaped nails.

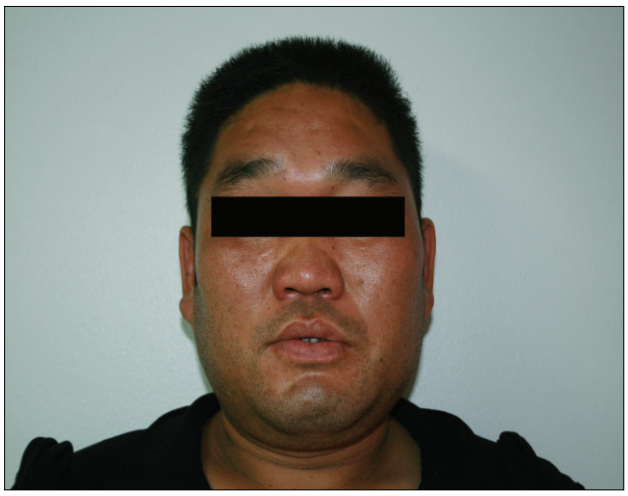

Fig. 1. Photograph of the patient's face shows deep frontal folds (furrowing) and oily facial skin.

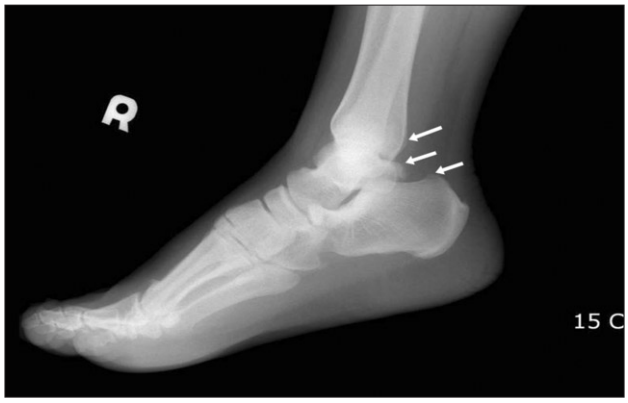

Fig. 3. The X-ray of right lower leg demonstrates irregular outline and periosteal new bone formation of the calcaneus and talus bone (lower and middle arrows). Also, periosteal new bone formation is at the distal right tibia (upper arrow).

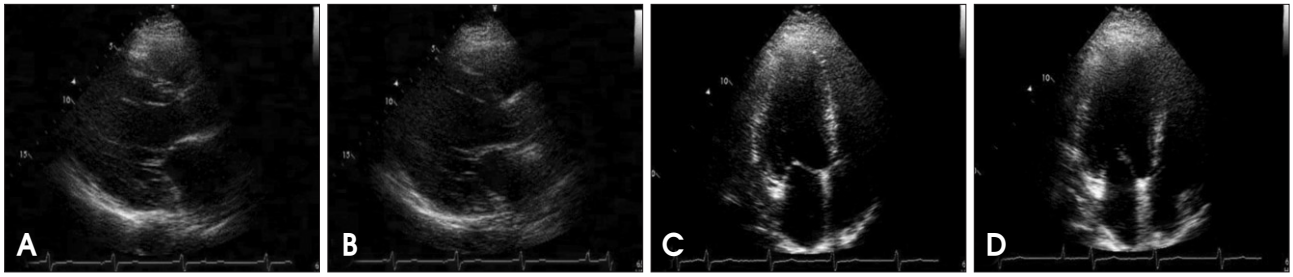

Fig. 4. Two-dimensional echocardiography on admission. Parasternal long axis view (A: end-systolic, B:end-diastolic) and apical 4 chamber view (C: end-systolic, D: end-diastolic) show eccentric left ventricular hypertrophy and enlarged left atrium.

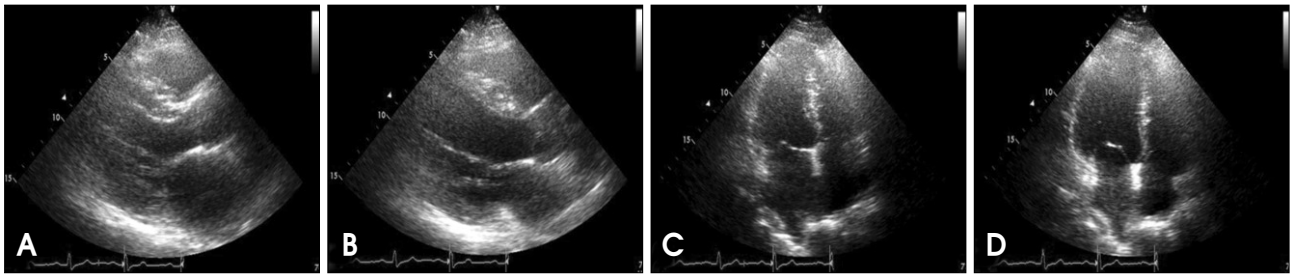

Fig. 5. Two-dimentional echocardiography on 3 months later. Parasternal long axis view (A: end-systolic, B: end-diastolic) and Apical 4 chamber view (C: end-systolic, D: end-diastolic) show normalized left ventricular internal diameter compared with that on admission (Fig. 4).

nary congestion and a disappearance of dyspnea. After discharge, diuretics and ACE-I maintained, beta-blocker, digoxin, nitrate and angiotensin receptor blocker were added.

After 3 months of treatment for heart failure, TTE showed normalized LV chamber dimensions (LV end-diastole dimension=48.5 mm) and LV systolic function (LV EF= 64.8%) (Fig. 5).

Discussion

Pachydermoperiostosis was first reported in 1868 and it was then thought to be examples of acromegaly. The first to recognize this as a distinct entity from acromegaly or pulmonary hypertrophic osteoarthropathy was in 1935. Pachydermoperiostosis is considered to be hereditary, even though a family history of the disease can, in fact, only be traced in around 25% to 38% of cases.[2] The precise incidence of the disease is unknown.

The clinical manifestations are somewhat variable, with affected patients demonstrating either the complete syndrome (pachydermia, periostosis, clubbing), the incomplete form (no pachydermia), or the forme fruste (pachydermia with minimal or absent periostitis).[3]

A differential diagnosis is required given the clinical similarity to acromegaly, which is also accompanied by skin abnormalities, including cutis verticis gyrata. In the case of acromegaly, however, bones in general are larger in the face, jaw (prognathism), skull, and limbs, and this is very evident in a radiographic study in the absence of signs of periostosis.[1] The underlying pathogenic mechanism of this disease remains unclear. Numerous theories have been proposed for the pathogenesis of pachydermoperiostosis.

The hormonal theories include a possible role for steroids, cytokines, and growth factors.[4]

Our patient had the complete form of pachydermoperiostosis, since he had hyperostosis, finger clubbing, and pachydermia. A variety of associated abnormalities have been described such as cranial suture defects, female escutcheon, bone marrow failure, gastric ulcer, hypertrophic gastropathy and Crohn's disease as accompanying diseases.[5][6] Pachydermoperiostosis accompanied by heart failure has not been reported so far.

Most hypertensive patients on admission have LV remodeling on echocardiogram.[7] So it is possible that heart failure in this case is associated with hypertension. But, it is difficult to exclude the possibility that heart failure is associated with pachydermoperiostosis. In order to exclude this problem, further research will be necessary.

Most patients present with moderate pain and swelling in multiple joints. Treatment, which is symptomatic and aim at attenuating the bone pain, is based on nonsteroidal anti-inflammatory drugs, pamidronate, or colchicines.[2] Fortunately, our patient has no complaints with joint pain and other symptoms. So, our decision to treatment with pachydermoperiostosis is follow-up and observation.

We conclude that a diagnosis of pachydermoperiostosis requires a high degree of clinical suspicion, given that we come near to misdiagnose as acromegalic cardiomyopathy. We hope this case help other physicians to diagnose pachydermoperiostosis accompanying heart failure correctly.

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