

A Case of Hereditary Transthyretin Cardiac Amyloidosis Presenting Complete Atrioventricular Block

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Background: Hereditary transthyretin (TTR)-related amyloidosis (mATTR) is an autosomal dominant disease that causes the extracellular deposit of misfolded TTR. The p.Asp58Ala (c.173A>C) is the most common mutation in Korea. The involvement of the cardiac conduction system is frequently observed but, the initial presentation of the complete atrioventricular block (AVB) without heart failure is uncommon. Here, we report a case of mATTR patient initially presenting with syncope.

Case: A 63-year-old man without any medical history was visited in the emergency room with syncope. His ECG revealed a complete AVB (Figure 1). He underwent permanent pacemaker implantation. His transthoracic echocardiography showed thickened left ventricular walls with high echogenicity and normal function. (Figure 2). The E velocity was 124 cm/s, e prime velocity 3.1 cm/s, and E/e' ratio 31, and the inferior vena cava (IVC) plethora was also observed, suggesting elevated both atrial pressure. These findings mean infiltrative cardiomyopathy such as cardiac amyloidosis. His protein electrophoresis and immunofixation electrophoresis of the light chain was negative, and a 99mTc-3,3-diphosphono-1,2-propanodicarboxylic acid (DPD) bone scan showed grade III uptake in the myocardium (Figure 3). Troponin I and brain natriuretic peptide was 112 pg/ml (normal range < 60) and 143 pg/ml (normal range < 39), respectively. In an endomyocardial biopsy, Congo-red staining revealed scarlet-colored amorphous materials, which show apple-green birefringence under polarized light (Figure 4). Serum TTR gene mutation test confirmed the p.Asp58Ala point mutation. Collectively, he was diagnosed with mATTR. He denied any neurologic symptoms, but on the study of nerve conduction velocity, spinal stenosis and carpal tunnel syndrome were observed.

Discussion: mATTR can present various manifestations of neurologic and cardiac involvements. This case showed an initial presentation of complete AVB without any heart failure symptoms; thus, high suspicion was needed. A comprehensive assessment of echocardiography can provide a critical clue to diagnosis, and a 99-Tc DPD bone scan is helpful to confirm the diagnosis.

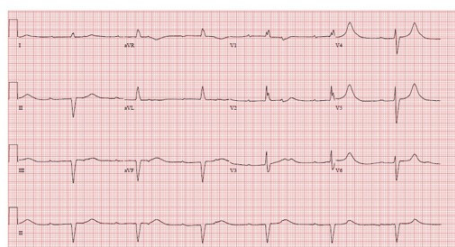


Figure 1) ECG, Complete atrioventricular (AV) block



Figure 2) TTE showed thickened left ventricular walls with high echogenicity.

99mTc-DPD Whole Body Bone Scan

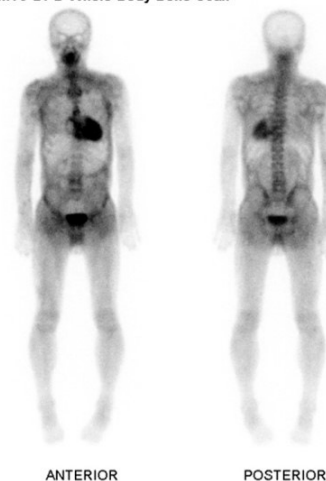


Figure 3) 99mTc-DPD bone scan showed strong uptake in the myocardium. (grade III)

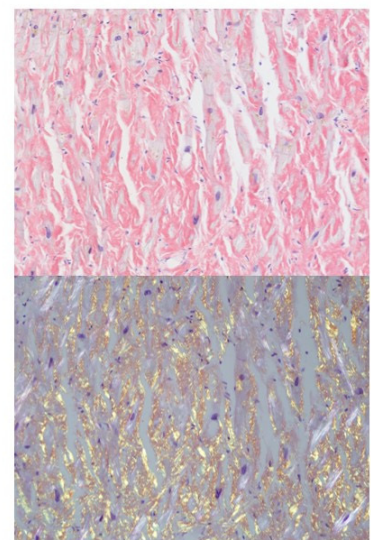


Figure 4) Congo red stain, optical microscope (▲) revealed scarlet-colored amorphous material and polarized light microscope (▼) show apple-green Birefringence.