

A Case of Arrhythmogenic Right Ventricular Cardiomyopathy in Patient with Type 1 Myotonic Dystrophy

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Type 1 myotonic dystrophy (DM1) is associated with a variety of cardiac conduction abnormalities and myopathy. There is several evidences from previous literatures that arrhythmogenic right ventricular cardiomyopathy (ARVC) is due to mutations in the dystrophin myotonic protein kinase (DMPK) gene, which have also been implicated in DM1 but coincidence of ARVC and DM1 is extremely rare. A 28-year-old man presented with ventricular fibrillation cardiac arrest and resuscitated successfully. The patient had no known comorbidities. In the patient's normal sinus rhythm electrocardiography, epsilon waves were observed (Figure 1A), and on the transthoracic echocardiography, structural and functional abnormalities consistent with ARVC in right ventricle were identified (Figure 1B). ARVC was diagnosed based on two major criteria. Through evaluation of the family history physical examinations, he was also suspected to have a neuromuscular disease. He was diagnosed with DM1 by genetic test and had >150 number of cytosine-thymine-guanine (CTG) repeats. However, the most common genes reported in ARVC, plakophilin-2 (PKP2), and desmoglein-2 (DSG2) were not identified in the patient. Since the patient fulfilled a number of the Task Force Criteria for ARVC, the patient was diagnosed with both DM1 and ARVC. The patient underwent implantable cardioverter defibrillator (ICD) placement as a secondary prevention and discharged after recovery and rehabilitation. To our knowledge, this case report is a very rare presentation of coincidence of DM1 and ARVC. Indeed, given that DM1 patients with a higher number of CTG repeats are more likely to have cardiac involvement, screening for ARVC could be crucial

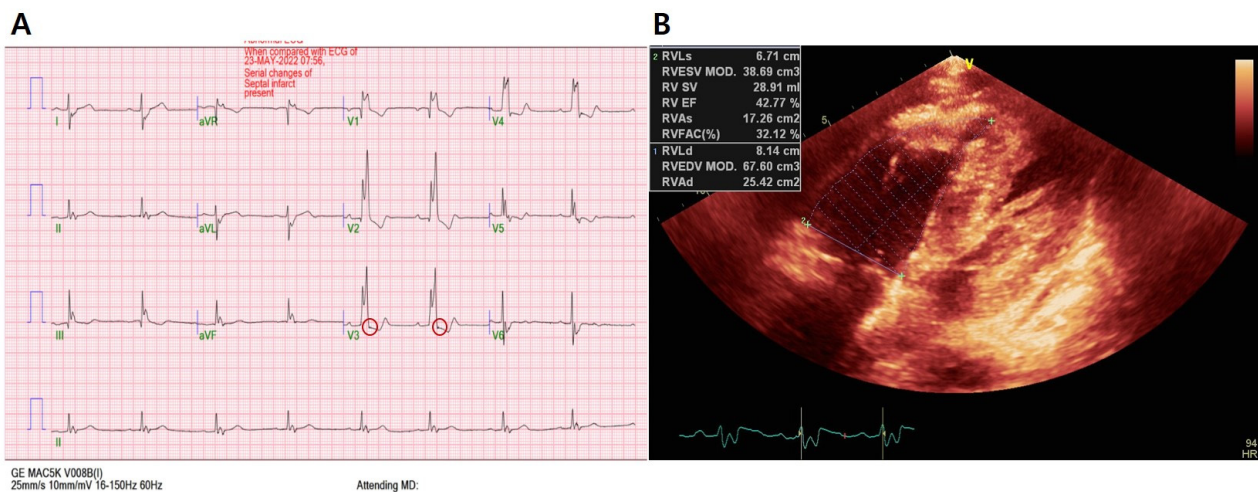


Figure 1. A. Reproducible epsilon waves in precordial V3 lead, B. Regional RV dyskinesia and RV fractional area change $\leq 33\%$.