Case Report

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Intermittent Pre-Excitation-Syndrome in Facio-Scapulo-Humeral Muscular Dystrophy

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Pre-excitation-syndrome has not been reported as a phenotypic feature of facio-scapulo-humeral muscular dystrophy (FSH-MD). In a 39-year-old male with FSH-MD due to a reduced tandem repeat size in the D4Z4-locus on chromosome 4q35, cardiac involvement, manifesting as an incomplete right bundle-branch-block, tall T-waves in V 3-5, ST-elevation in V 2-4, and mild thickening of the left ventricular myocardium, was first recognised 10 years earlier. Follow-up at age 39 years revealed mild myocardial thickening, two intra-ventricular aberrant bands, and, surprisingly, intermittent pre-excitation on a routine electrocardiography. Cardiac involvement in FSH-MD may manifest as hypertrophic cardiomyopathy or various arrhythmias, of which one may be pre-excitation-syndrome. **(Korean Circ J 2014;44(5):348-350)**

KEY WORDS: Heart; Muscular dystrophy, Fasuoscapulohumerales; Arrhythmia; Pre-excitation syndromes; Cardiomyopathies.

Introduction

Though occasionally reported, cardiac involvement is a rare phenotypic feature of facio-scapulo-humeral muscular dystrophy (FSH-MD).[1)] Cardiac involvement in FSH-MD includes cardiomyopathy and arrhythmias.[1)] Though various arrhythmias have been found in patients with FSH-MD, pre-excitation-syndrome has not been reported.

Case

The patient is a 39-year-old veterinarian who developed slowly progressive muscle weakness and wasting of the shoulder girdle muscles since age 3 years. Initial diagnostic work-up, including muscle biopsy, was non-informative. Since age 29 years he additionally developed diffuse weakness and wasting of the left leg accompanied by muscle stiffness. Since then, he also noted generalized muscle tenderness, muscle aching after exercise, and contractures. Creatine-kinase (CK) was repeatedly mildly elevated. Needle-electromyography was myogenic and deoxyribonucleic acid analysis revealed a reduction of the FSH-MD tandem repeat size in the D4Z4 locus on chromosome 4q35 (Fig. 1). Based upon the clinical and genetic findings, FSH-MD was diagnosed at age 30 years. The family history was negative for primary myopathy.

Follow-up investigation in July 2012 revealed a facies myopathica with weakness of the upper lids, inability to voluntarily balloon the cheeks, and weak anteflexion of the head (M5-). There was prominent scapular winging, bilateral diffuse weakness of the upper limbs with right-sided predominance, hypotonia, diffuse wasting, and reduced deep tendon reflexes. On the lower limbs, there was bilateral weakness of hip flexion (M4) and foot extension (M4-). Patella tendon reflexes were preserved but Achilles tendon reflexes were reduced. There was asymmetric wasting with left-sided predominance, bilateral hypotonia, lumbar hyperlordosis, and a waddling gait. Nevertheless, he was able to play the guitar and drive an automatic car, even over long distances. Cardiologic investigation at follow-up revealed mild myocardial thickening, two intra-ventricular aberrant bands, and, surprisingly, intermittent pre-excitation on routine

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electrocardiography (Fig. 2). Stress testing to see if there was predominance of prolonged QRS-complexes and thus indication for ablation or predominance of normally-sized QRS-complexes, clearly demonstrated a reduction of prolonged QRS-complexes. Since he was asymptomatic and pre-excitation-syndrome occurred only intermittently, he was not recommended to undergo ablation.

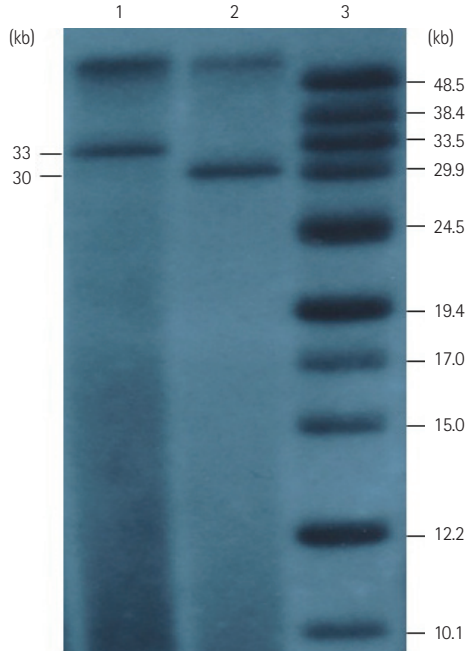

Fig. 1. Hybridization of a southern blot of the patient's DNA with radioactively labelled probe p13E-11 (D4F104S1); lane 1: DNA cleaved by *Eco* RI, lane 2: DNA cleaved by *Eco* RI+Bln I, lane 3: Lambda Mix Marker, 19 (Thermo Scientific Molecular Biology).

Discussion

The presented case is interesting for the association of pre-excitation syndrome and FSH-MD. Rhythm abnormalities so far reported in FSH-MD include impulse generation abnormalities, such as sinus node dysfunction.[2)3)] supraventricular arrhythmia,[3)4)] and bradycardia,[3)4)6-8)] or impulse propagation abnormalities, such as short PR-interval,[4)] tall P-waves,[2)3)] abnormal atrio-ventricular conduction with complete atrio-ventricular block,[3)4)] abnormal Q-waves,[8)] intraventricular conduction delay,[3)4)8)] incomplete bundle branch block,[2)] ventricular tachycardia,[3)] abnormal ST-segment,[3)] high T-waves,[3)4)] prolonged QT-interval,[6)] increased R/S-ratio in V1,[7)] or hypertrophy.[2)] In a study of 83 patients with FSH-MD, 12% had cardiac arrhythmias.[4)] In a study of 24 patients with FSH-MD, 9 had positive ventricular late potentials and increased QT-dispersion compared to controls.[9)] In the two patients described by Laforêt et al.[4)] a shortened PR-interval was not associated with a delta-wave.

Discrete concentric thickening of the myocardium was also interpreted as cardiac involvement in the presented case, since the history was negative for arterial hypertension or a thoracic deformity. Single patients with FSH-MD also develop cardiomyopathy.[10)]

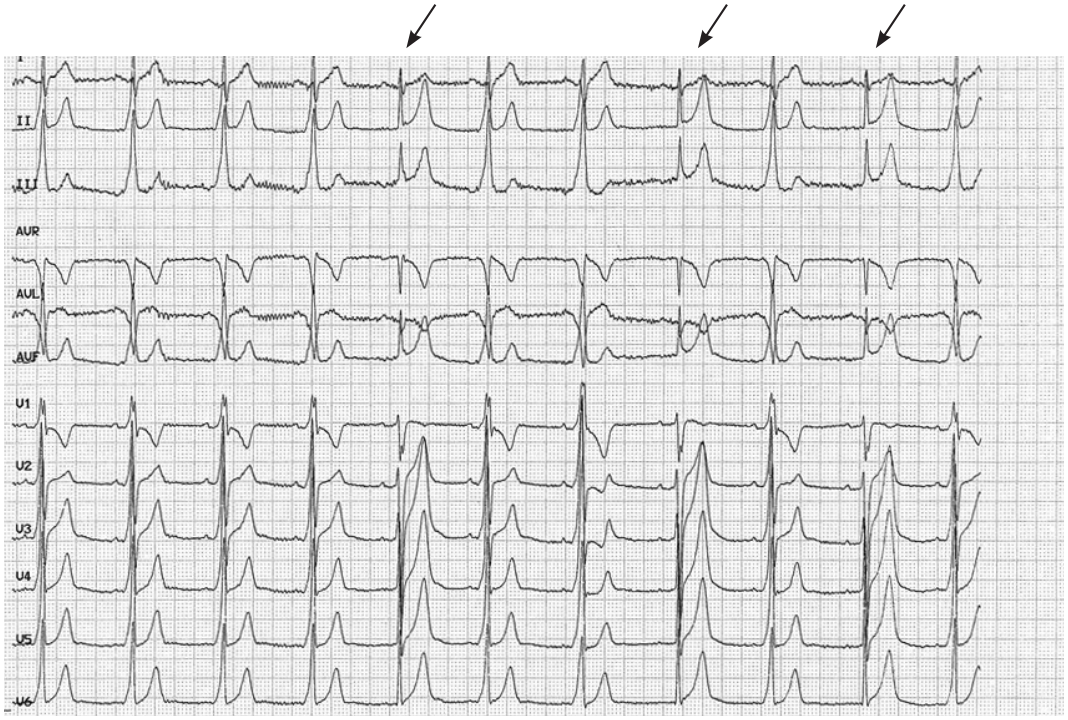

Fig. 2. Routine electrocardiography showing intermittent pre-excitation-syndrome interrupted by QRS-complexes without delta-wave. Three beats are normally conducted (arrows).

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Accordingly, echocardiography in FSH-MD may show enlarged right cardiac cavities or restricted right ventricular movement, attributed to thoracic deformities[2)8)] or hypertrophic cardiomyopathy.[4)10)] Additionally, myocardial scintigraphy revealed reduced Thallium-201 uptake due to suspected myocardial fibrosis.[7)]

This case shows that there is indeed cardiac involvement in FSH-MD, manifesting either as hypertrophic cardiomyopathy or arrhythmias. Among various different arrhythmias described in FSH-MD, single patients may also develop pre-excitation-syndrome.

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