LEFT VENTRICULAR NONCOMPACTION CARDIOMYOPATHY: ADULT ASSOCIATION WITH 1p36 DELETION SYNDROME

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A 25-year-old patient with a 1p36 gene deletion was referred to cardiac magnetic resonance (CMR) imaging for evaluation of left ventricular noncompaction cardiomyopathy (LVNC). She had a recent history of palpitations that were not associated with chest pain or syncope. Electrocardiogram showed left ventricular hypertrophy with a strain pattern (Figure 1 H). Holter monitor showed only rare premature ventricular contractions. CMR showed prominent left ventricular trabeculation (Figure 1 A-E). The end diastolic compacted myocardium to noncompacted myocardium ratio met the CMR diagnostic criteria of > 2.3 for

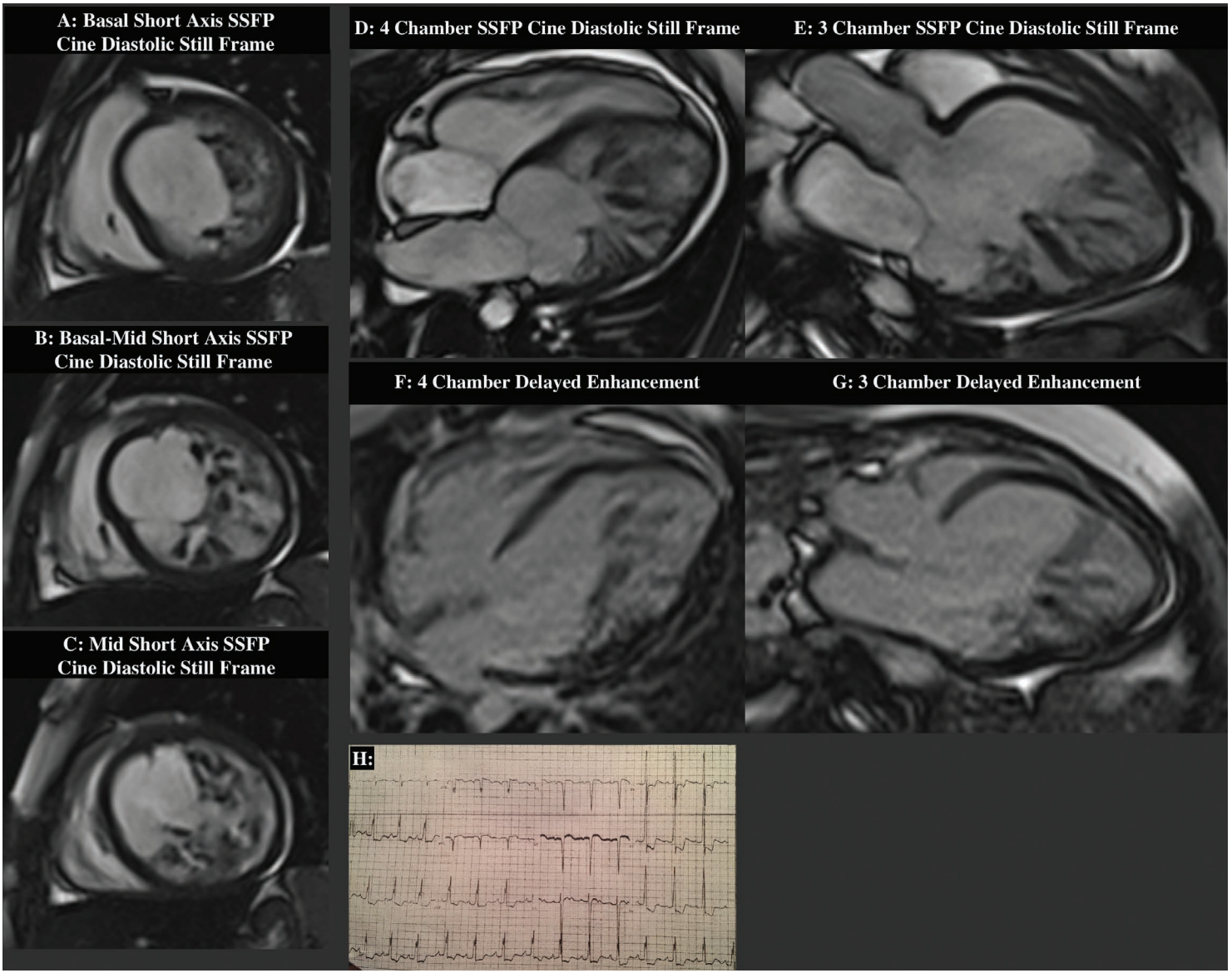

Figure 1. Electrocardiogram (EKG) images of patient with left ventricular noncompaction cardiomyopathy. (A-E) Three short-axis, 4-chamber, and 3-chamber SSFP cine still frames showing increased trabeculation, meeting magnetic resonance imaging criteria for noncompaction cardiomyopathy. (F, G) Delayed-contrast enhanced images with high inversion time (T1600) showing no thrombus in noncompacted tissue. (H) EKG consistent with left ventricular hypertrophy with strain pattern.

LVNC. No scar or thrombus was seen within the trabeculation on delayed contrast images (Figure 1 F-G). The patient was referred to electrophysiology for consideration of implantable cardiac defibrillator (ICD).

LVNC is a well-described cause of cardiomyopathy, and current criteria rely on echocardiography for diagnosis.[1] CMR imaging is particularly useful for the diagnosis of LVNC.[2] As LVNC is more commonly recognized, the identification of a genetic basis becomes increasingly important. LVNC has been described in a number of genetic syndromes, including 1p36.[3] The development of the 1p36 gene deletion is a microdeletion syndrome that is a common cause of developmental delay and mental retardation, with a frequency of approximately 1 in 5,000 newborns.[4] In a series of 60 patients with 1p36, Battaglia et al. described a 23% frequency of LVNC.[5] Recently, the terminal 14 exons of the transcription factor PRDM16 in the 1p36 region were found to cause cardiomyopathy in patients with 1p36, LVNC, and dilated cardiomyopathy.[6]

We present, to the best of our knowledge, the first case of LVNC associated with a 1p36 deletion in an adult evaluated with CMR imaging. The role of ICD for primary prevention in this population is not fully delineated and requires further investigation.

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Keywords: left ventricular noncompaction cardiomyopathy, cardiomyopathy, 1p36 gene deletion, microdeletion syndrome

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