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IMAGES AND VIGNETTES IN CLINICAL ELECTROPHYSIOLOGY

Multifocal Ectopic Purkinje-Related Premature Contractions Syndrome in R222Q-SCN5A Gene Mutation Carriers Treated With Flecainide

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A 21-year-old male patient with syncopal episodes and his 56-year-old father, with a family history of sudden cardiac death and dilated cardiomyopathy, were referred to our center. Physical examination and laboratory investigations were normal. The father's ambulatory electrocardiographic (ECG) abnormalities (**Figures 1A and 1B**) were poorly responsive to beta-blockers. The father's cardiac imaging showed mild left ventricular dilation and systolic dysfunction with no myocardial fibrosis. Catheter ablation of the right ventricle outflow tract (RVOT) was attempted, because of the prevalence of premature ventricle contractions at this level, and a regression of systolic dysfunction and dilation was observed (**Figure 2**), but no beneficial effect on the arrhythmic burden was obtained. Genetic testing was performed, revealing the R222Q-SCN5A gene mutation in both father and son, accountable for the rare multifocal ectopic Purkinje-related premature contractions (MEPPC) syndrome.[1,2] Therefore, flecainide via oral route was started with complete normalization of ECG (**Figure 1C**).

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The authors attest they are in compliance with human studies committees and animal welfare regulations of the authors' institutions and Food and Drug Administration guidelines, including patient consent where appropriate. For more information, visit the Author Center.

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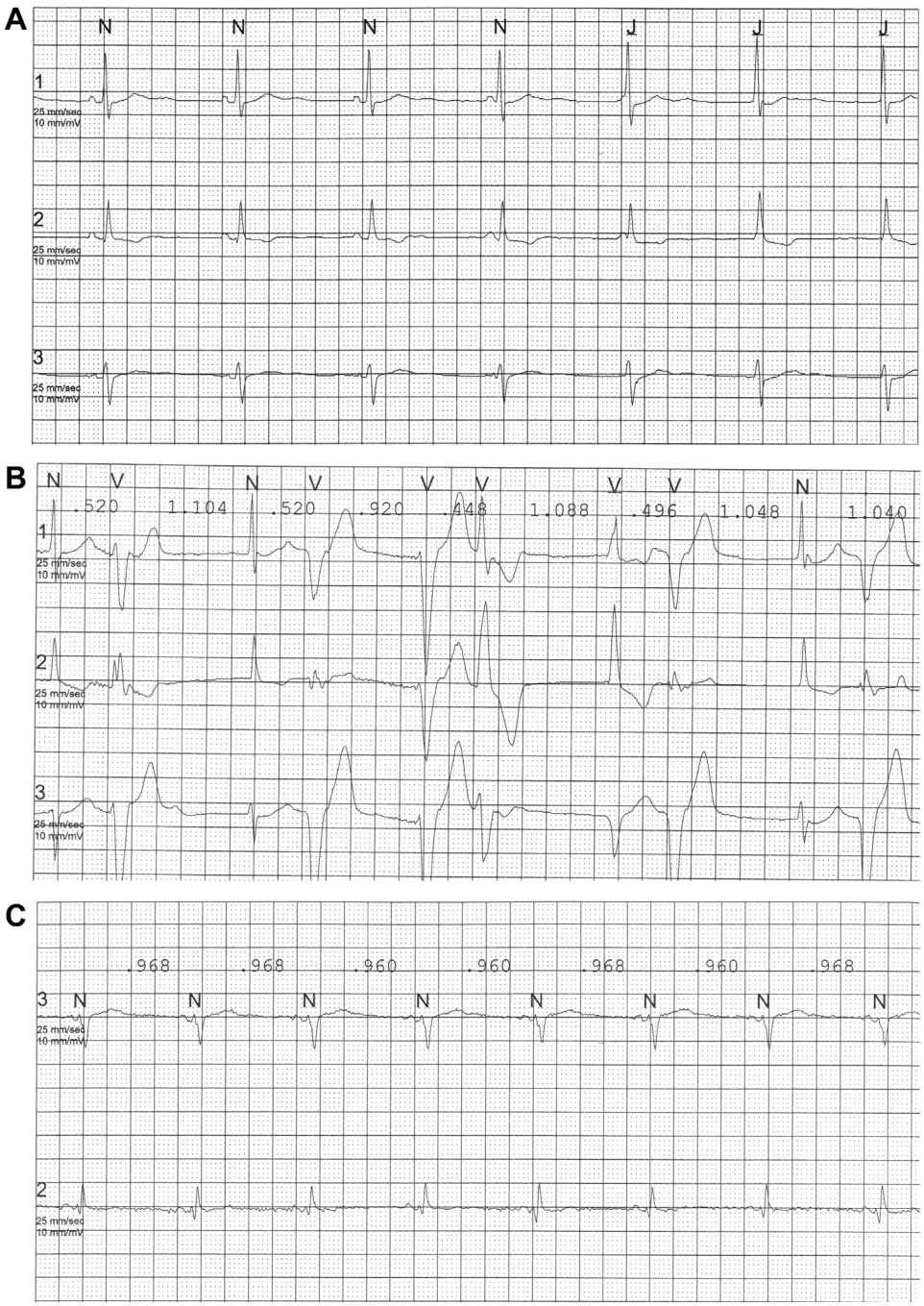
FIGURE 1 Father's Ambulatory Electrocardiogram Before and After Treatment With Flecainide



At baseline, the ambulatory electrocardiogram showed the typical electrocardiographic features of the multifocal ectopic Purkinje -related premature contractions (MEPPC syndrome): **(A)** junctional rhythm with isorhythmic atrioventricular dissociation; **(B)** premature ventricular contractions (PVCs) and nonsustained polymorphic ventricular tachycardia (NSVT). **(C)** After treatment with flecainide, normalization of the electrocardiogram was achieved with the abolition of PVCs and NSVTs. J = junctional complex; N = sinusal complex; V = ventricular complex.

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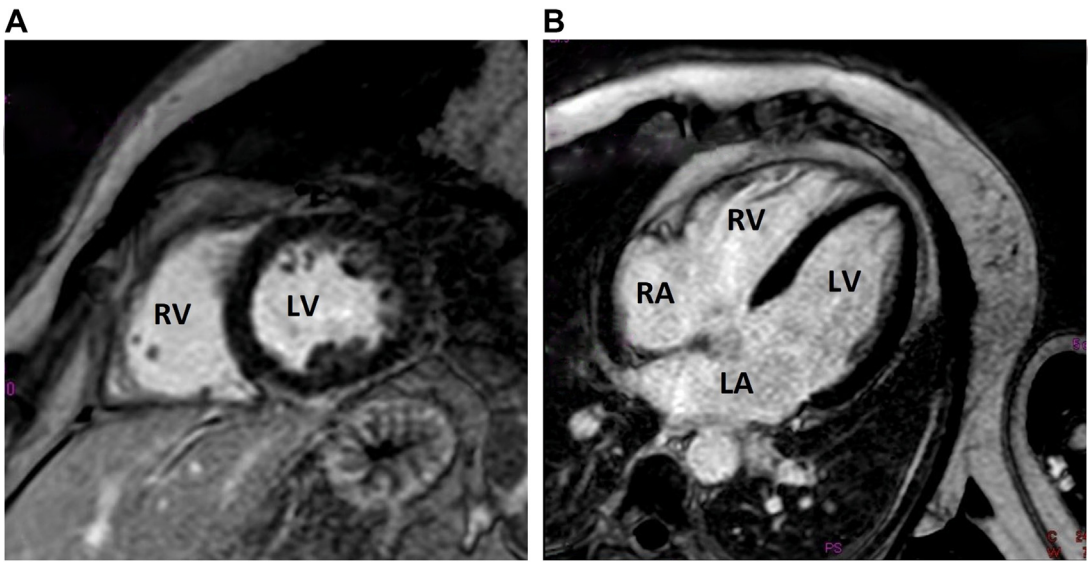
FIGURE 2 The Father's CMR Imaging, Showing the Absence of LV Dilation With a Normal Systolic Function, Without Myocardial Fibrosis at Late Gadolinium Enhancement Sequences



(A) Short-axis view at the level of papillary muscles. **(B)** Four-chamber view. LV = left ventricle; LA = left atrium; RV = right ventricle; RA = right atrium.

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KEY WORDS flecainide, MEPPC syndrome, SCN5A gene, ventricular arrhythmias