A Case of Non-Ischemic Cardiomyopathy

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Case Presentation:

A 25-year-old Caucasian landscaper with no significant past cardiovascular history, presented with recurrent sudden onset palpitations and blurred vision for few weeks. Family history was strongly positive for congestive heart failure.

Physical examination was rather unremarkable. A 12 lead EKG showed sinus rhythm, short PR interval (<120 ms), and a right bundle branch block (Figure 1). Laboratory work-up hs-troponin at 25 pg/mL (normal=<20) with minimal delta. Transthoracic echocardiogram findings were significant for mildly dilated, severely diffuse hypokinetic Left Ventricle with an EF of 20-25% and grade III diastolic dysfunction with elevated left atrial pressure, mild-moderate mitral and tricuspid valve regurgitation. Coronary CTA did not show coronary artery disease or anomaly. A cardiac MRI showed a LVEF of 38%, and RVEF of 30%, with no evidence of prior infarction or late gadolinium enhancement. He was sent home with maximum tolerable guideline-directed medical therapy for CHF and Wearable Cardiac Defibrillator.

Genetic panel for cardiomyopathy revealed heterozygosity for the LMNA gene mutation, supporting highly suspected diagnosis of Lamin A/C cardiomyopathy. He was found to have recurrent non-sustained VT on WCD (Figure 2-4). He did not have any syncopal events nor received shocks from wearable cardiac defibrillator. He received an AICD for primary prevention while genetic cascade testing for biologic family members being followed.

Discussion:

The prevalence of non-ischemic cardiomyopathy is estimated at 1/500 to 1/250 individuals (1). It predominantly affects younger individuals, while genetic correlations can be identified in about 40% of cases (2). Lamin A/C gene, is one of the most thoroughly investigated cardiomyopathy-related genes, which is associated with a higher incidence of Ventricular Arrhythmias or Sudden Cardiac death (SCD). Some studies suggest that the rate of SCD in Lamin A/C gene mutation carriers is as high as 46% (3, 4). In a retrospective study of 49 families of patients with dilated cardiomyopathy in Colorado, USA, and Italy, 12 carriers of the LMNA mutation were identified in younger patients (age of onset 27+/-5 years) compared to without the mutation, and their phenotype was frequently associated with electrophysiological abnormalities, such as supraventricular arrhythmias and conduction disease. Compared to non-carriers, carriers of the LMNA mutation were 2.6 times more likely to suffer cardiovascular death, 3.4 times more likely to experience cardiovascular death or transplant, and 2.2 times more likely to have cardiovascular death, transplant, or major event (5). European Society of Cardiology recommends consideration for an cardioverter defibrillator in patients with dilated cardiomyopathy and a confirmed disease-causing LMNA mutation with clinical risk factors [NSVT during ambulatory electrocardiogram monitoring, LVEF <45% at first evaluation, male sex and non-missense mutations (insertion, deletion, truncations or mutations affecting splicing)] (6).

Conclusion:

In younger patients with a strong family history of heart failure and early death, with minimal suspicion for drug-induced or infiltrative cardiomyopathies, genetic testing aids in the accurate

diagnosis, and guides for timely interventions for life saving therapies such as cardiac defibrillator. There is no need for 90 day waiting for such patients as despite improvement in EF, life threatening VA's are common with LMNA mutations.

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Figure Legend:

Figure 1. Base line ECG (NSR, Short PR interval and RBBB with repolarization changes)**,**Wide complex tachycardia on Wearable Cardiac Defibrillator**,** Cardiac MRI showing short axis images sowing no late gadolinium enhancement, Cross-Section of images**.**

Figure 2: Pedigree Tree **–** Of the last 3 generations

Figure 1:

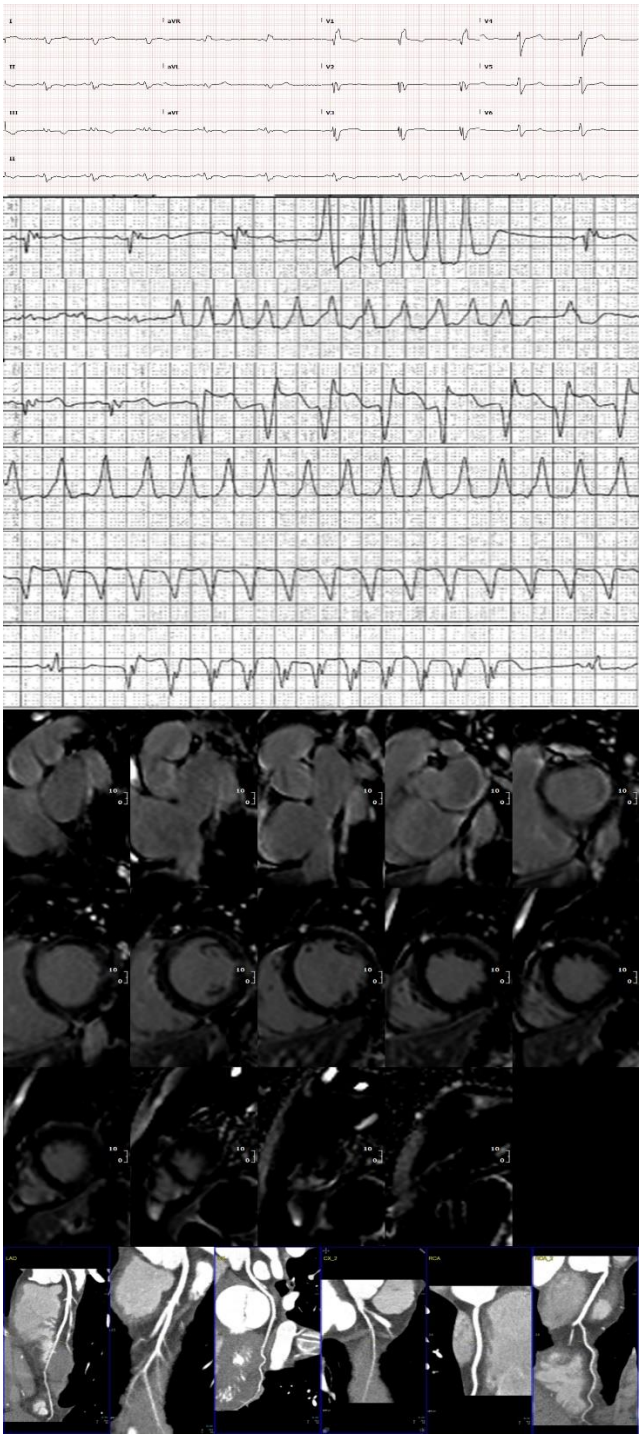
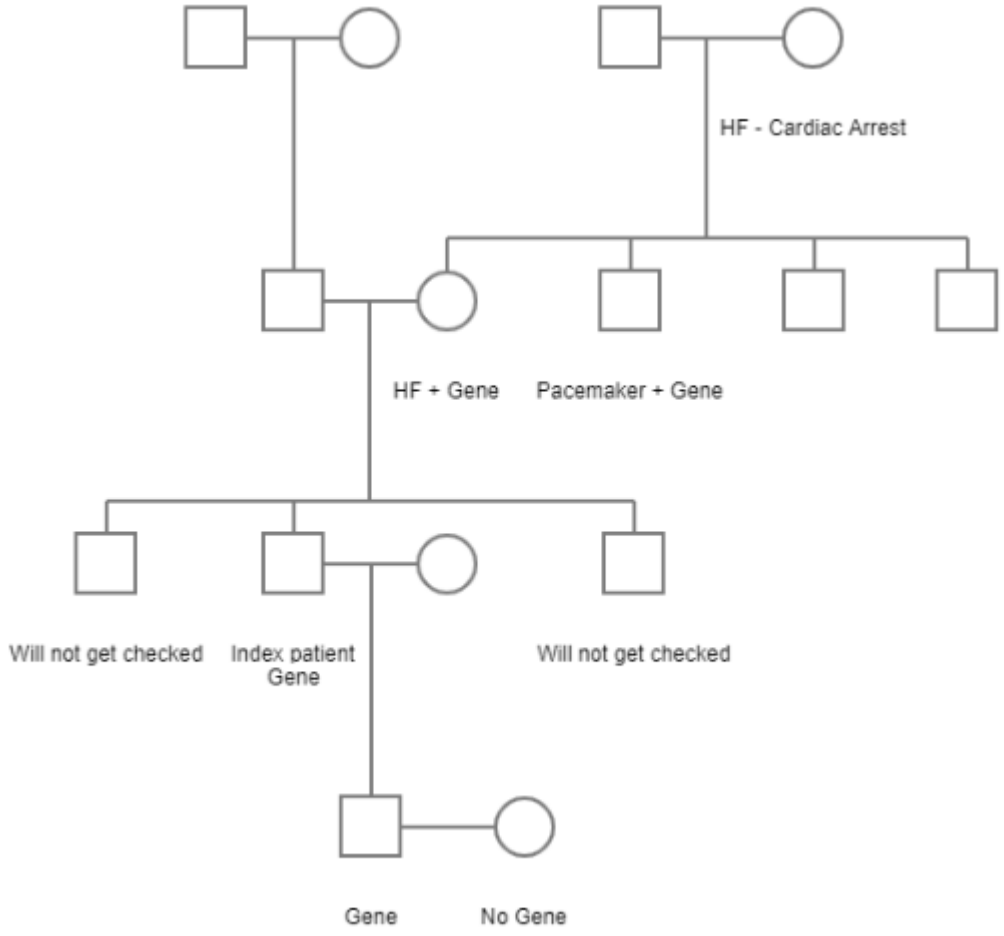


Figure 2:


HF - Cardiac Arrest
HF + Gene
Pacemaker + Gene
Will not get checked
Index patient
Gene
Will not get checked
Gene
No Gene