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References

Incremental value of three-dimensional strain imaging in Danon disease

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A 43-year-old woman with a familial history of Danon disease and early sudden death was clinically followed because of carrier of LAMP-2 mutation. She was symptomatic for palpitations. Clinical evaluation and electrocardiogram were normal (Panel A), while Holter monitoring revealed non-sustained ventricular tachycardia (VT; Panel B). Conventional two- and three-dimensional echocardiograms were unremarkable (see Supplementary data online, Movie S1); however, three-dimensional speckle-tracking imaging (3D-STI) revealed an impairment of the longitudinal strain of the basal segment of the inferior wall (blue area of the bull’s eye in Panel C). Contrast-enhanced cardiac magnetic resonance showed late gadolinium enhancement in the same area, indicating the presence of myocardial fibrosis (Panel D). Based on these findings and clinical data, the patient subsequently underwent implantable cardioverter-defibrillator (ICD) implantation and, shortly after, she received an appropriate ICD intervention for sustained VT.

Danon disease is a rare X-linked systemic disorder due to lysosome dysfunction affecting mainly young males; it is characterized by hypertrophic cardiomyopathy, skeletal myopathy, and mental retardation, leading to rapid clinical deterioration and death. LAMP-2 mutation results in profound myocardial structural abnormalities with extensive scarring replacement and autophagic and vacuolated myocytes; these may lead to life-threatening arrhythmias. Different phenotype expressions have been described in females ranging from asymptomatic carriers to hypertrophic or dilated cardiomyopathy with severe arrhythmogenic trait.

This case illustrates the potential role of new echocardiographic techniques for myocardial strain evaluation in risk stratification of female LAMP-2 mutation carriers. Three-dimensional STI may permit early recognition of cardiac involvement, through the detection of concealed myocardial abnormalities related to the presence of fibrosis, and guide ICD implantation.

Supplementary data are available at European Heart Journal – Cardiovascular Imaging online.