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Atrial standstill phenomenon in non-dilated cardiomyopathy phenotype of genetically confirmed laminopathy

Short title: Atrial standstill phenomenon connected to laminopathy

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Laminopathies as hereditary diseases may present different phenotypes. We described a case history of patient with LMNA laminopathy, without signs of left ventricular (LV) dilatation but with unusual atrial cardiomyopathy and severe right ventricle dilatation.

A 57-years old man diagnosed with LMNA cardiomyopathy was hospitalized due to the progression of heart failure (HF) signs and symptoms. His medical history onset was in 2013 (Figure 1A), when at age 44 he was diagnosed with atrial fibrillation (AF) and sick sinus syndrome, underwent pulmonary vein isolation connected with left atrial appendage closure, followed by the dual chambers pacemaker implantation. Regardless of atrial appendage closure anticoagulation treatment was maintained. Because of relatively young age and unexplained conduction disorders he was referred to the genetic counseling and in genetic test was positive for LMNA laminopathy — variant NM_001282625.1:p.Gln6*/c.16C>T. This mutation was observed in Emery–Dreifuss dystrophy.

In the following years the conduction disturbances were in progress finally leading to the third-degree atrioventricular block. Regardless of the lack of HF symptoms taking into regard

the baseline diagnosis and the need of persistent stimulation the pacemaker was up-graded into cardiac resynchronization therapy defibrillator (CRT-D).

For the next 6 years patient did not experience any deterioration. After that time, during hospitalization because of CRT-D battery depletion, the following abnormalities were observed: HF decompensation, AF, increased N-terminal pro-B-type natriuretic peptide concentration (442 pg/ml), worsening of LV ejection fraction (LVEF: 40%, normal LV dimensions, E/e' = 15, LA 44 mm) were found. However, the detailed ECG re-analysis (Figure 1B) did not confirm AF, but the atrial stimulus with no atrial response was present. The CRT-D control (Figure 1D) showed that there is no atrial electrical activity as well as no conduction even after increasing threshold to maximum.

Because of the significant right ventricle enlargement and signs of pulmonary hypertension (PH) (Figure 1C) the right heart catheterization was performed and the post-capillary PH was diagnosed (mean pulmonary pressure: 27 mm Hg, PAWP: 20 mm Hg, pulmonary vascular resistance: 1,9 Wood units). Patient was discharged with typical HF treatment (bisoprolol 5mg/d), anticoagulation (dabigatran 2 × 150 mg/d) and sent to strict ambulatory control.

Summarizing, the presented case is an example of "non-dilated" cardiomyopathy phenotype related to the LMNA laminopathy. We should be aware that both AF and conduction disturbances in the young subject may be the first presentation of genetic disorder. Thus, the detailed diagnostic process including genetic tests is necessary and may improve prognosis for these patients. When non-dilated LV cardiomyopathy phenotype is recognized it is recommended to test the same gene panel as in dilated cardiomyopathy [1].

Special interest should be put on the lack of atrial electrical activity — this is a rare phenomenon, called atrial standstill, especially connected with laminopathy diagnosis. Furthermore, authors described this phenomenon with accompanying multiple arterial embolism [2]. The reason of an elevated risk of arterial embolism is lack of atria conduction and almost no contractility function. To answer the question when AF turned into atrial standstill the electrophysiology study should have been done.

It has been confirmed [3] that novel pathogenic variant of LMNA gene could manifest as atrial cardiomyopathy. Moreover, it has been proved that LMNA mutation and elevated N-terminal pro-B-type natriuretic peptide concentration is connected with increased mortality [4]. It additionally argues that our patient requires strict observation and multidirectional therapy. Atrial standstill and postcapillary PH as a result of HF with mildly reduced EF, diastolic dysfunction and atrial remodeling are indirect markers of the disease advancement.

Article information

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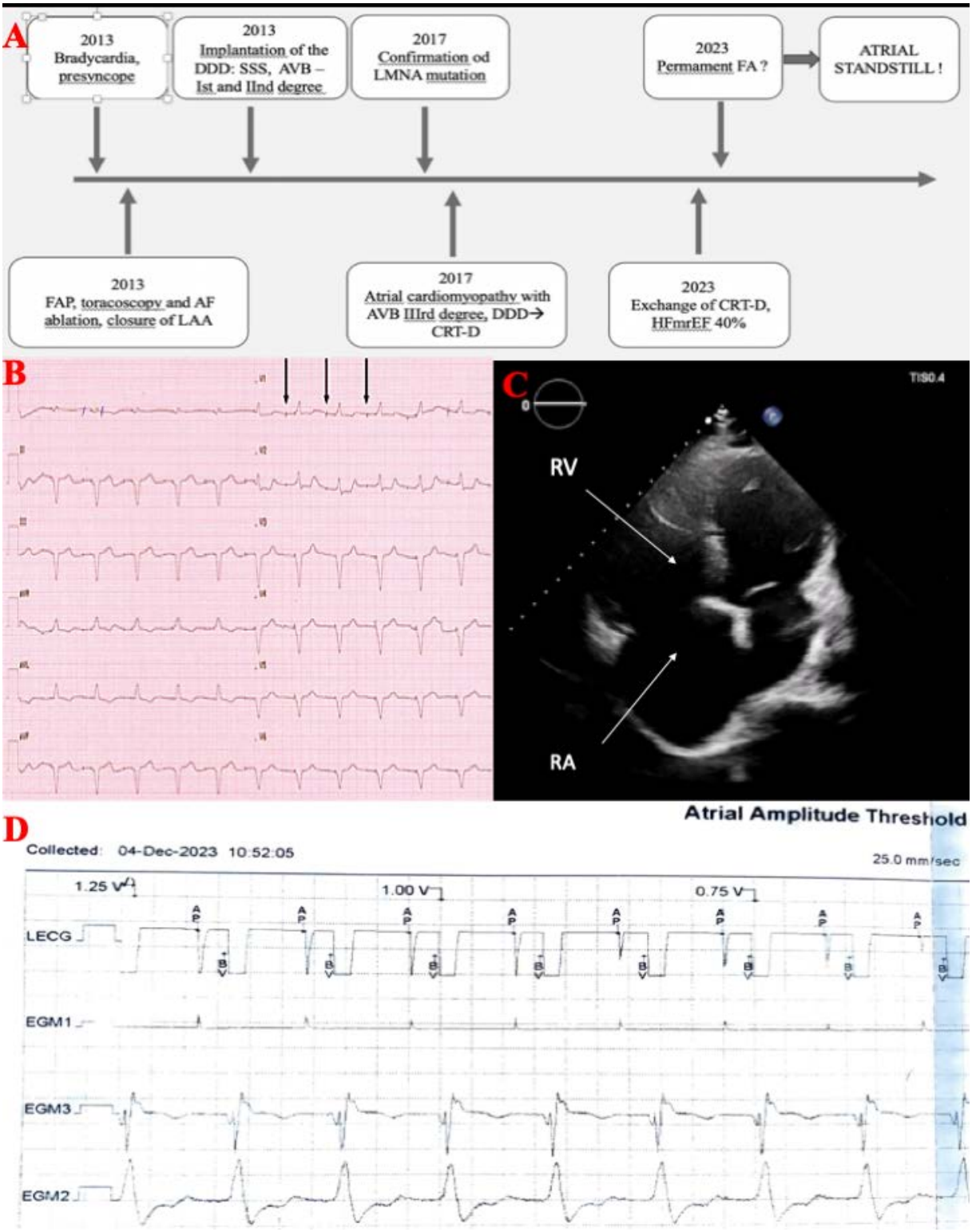

Figure 1. A. Flowchart. **B.** EKG — atrial stimulation without any response, BIV stimulation. **C.** Transthoracic echocardiography — 4-chamber view. Enlargement of right ventricle and right atrium. **D.** CRT-D record – atrial stimulus with no response