I. HISTORY OF PHOSPHOLAMBAN MUTATION IN ARRHYTHMOGENIC CARDIOMYOPATHY: A CASE REPORT

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CASE REPORTS

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Abstract: Phospholamban mutations represent a very special form of arrhythmogenic cardiomyopathy with the risk of developing heart failure in middle-aged patients. A special history of a phospholamban mutation of the female patient is described. Although arrhythmogenic left dominant ventricular cardiomyopathy is suggestive, distinctive findings of MRI imagiing techniques and standard ECG predict typical appearance of phospholamban cardiomyopathy.

Keywords: phospholamban; arrhythmogenic cardiomyopathy; inferolateral T wave inversions; low voltage in limb leads

Introduction.

Here a case report is presented of a 60 year old female patient, whose sister was phospholamban positive with a therapy resistant decompensated heart failure supplied with left and right ventricular assist device. Despite these arrangements the patients died.

The female patient was phospholamban positive, too. There were no complaints.

In 2015 cardiac MRI at the Heart and Diabetes Centre in Bad Oeynhausen, Germany, revealed borderline ventricular dimensions. Late enhancement was negative.

Repeated cardiac MRI in 2019 at the same institution showed dyskinesia of the right ventricular apex. Late enhancement was positive in the septal and inferoseptal regions of the left ventricle.

The ECG in 2020 revealed low voltage in limb leads and T wave inversions in inferolateral leads – the typical electrocardiographic sign of isolated arrhythmogenic left ventricular cardiomyopathy. The patient was treated prophylactically with eplenerone to reduce fibrosis and beta blocking agents.

Echocardiography revealed non dilated left ventricle with a slightly reduced ejection fraction of 50%. Right ventricle showed isolated dilatation of the right ventricular outflow tract and the inferior area. In the ECG low voltage in limb leads were present, additionally T-wave inversions in inferior and lateral leads from leads V3 up to V6.

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Discussion

The long-going history of a phospholamban-positive female patient is presented in this case report. The sister of the patient died despite left and right ventricular assist devices. Although the patient was phospholamban positive in 2015, cardiac MRI revealed borderline ventricular dimensions without positive late enhancement.

Four years later the patient had no complaints, but repeated cardiac MRI showed right ventricular outflow tract dyskinesia without evidence of fibrosis or fatty infiltation, but positive late enhancement on the apical and inferoseptal regions of the left ventricle.

In 2020 the standard ECG of the patient revealed typical signs of isolated arrhythmogenic left ventricular cardiomyopathy by low voltage in limb leads and inferolateral T-wave inversions.

In this case the history of the phospholamban mutation is strange. In normal circumstances phospholamban mutations progress in short term leading to therapy resistent heart failure (1) as described in the sister of the patient. An isolated arrhythmogenic left ventricular cardiomyopathy by MRI imaging and ECG (2) is extremely rare, not yet described in phospholamban mutations. The patients remains controlled by echocardiography and holter monitoring each year, control cardiac MRI is planned in again 4 years. Late enhancement in arrhythmogenic strict left ventricular cardiomyopathy due to mutatons in desmoplakin, desmoglein-2, desmin and other mutations is localised to posteroapical area, in phospholamban mutation in septal and inferoseptal locations.

The ECG of patients differ in T-wave inversion in lateral leads: strictly in leads V5 and V6 in arrhythmogenic left

ventricular cardiomyopathy, and globally in leads V3 or V4 up to V6 in phospholamban mutations.

Conflict of interests

There are conflicts of interest to declare

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