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CASE REPORT

Arrhythmogenic right ventricular dysplasia: Atypical clinical presentation☆

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Abstract A 67-year-old man was admitted to our hospital after episodes of syncope preceded by malaise and diffuse neck and chest discomfort. No family history of cardiac disease was reported. Laboratory workup was within normal limits, including D-dimers, serum troponin I and arterial blood gases. The electrocardiogram showed sinus rhythm with T-wave inversion in leads V1 to V3. Computed tomography angiography to investigate pulmonary embolism showed no abnormal findings. Transthoracic echocardiography (TTE) displayed massive enlargement of the right ventricle with intact interatrial septum and no pulmonary hypertension. Cardiac magnetic resonance imaging (MRI) confirmed right ventricular (RV) dilatation and revealed marked hypokinesia/akinesia of the lateral wall. Exercise stress testing was negative for ischemia.

According to the 2010 Task Force criteria for arrhythmogenic right ventricular dysplasia (ARVD), this patient presented two major criteria (global or regional dysfunction and structural alterations: by MRI, regional RV akinesia or dyskinesia or dyssynchronous RV contraction and RV ejection fraction $\leq$40%, and repolarization abnormalities: inverted T waves in right precordial leads [V1, V2, and V3]); and one minor criterion (>500 ventricular extrasystoles per 24 hours by Holter), and so a diagnosis of ARVD was made.

After electrophysiologic study (EPS) the patient received an implantable cardioverter-defibrillator (ICD).

This late clinical presentation of ARVD highlights the importance of TTE screening, possibly complemented by MRI. The associated risk of sudden death was assessed by EPS leading to the implantation of an ICD. Genetic association studies should be offered to the offspring of all ARVD patients.

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PALAVRAS-CHAVE
Displasia arritmogénica do ventrículo direito;
Ecocardiografia transtorácica;
Ressonância magnética cardíaca;
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Estudo eletrofisiológico cardíaco;
Cardioversor-desfibrilhador implantável

Miocardiopatia arritmogénica do ventrículo direito – particularidades de um caso

Resumo Um homem de 67 anos deu entrada no hospital após episódios de síncope precedidos por mal-estar geral e desconforto torácico difuso. Sem história familiar de doença cardíaca. O estudo analítico, incluindo d-dímeros e troponina I, bem como a gasimetria arterial, não mostrou alterações. O eletrocardiograma exibiu um ritmo sinusal com inversão da onda T de V1-V3. A angioTC foi negativa para tromboembolismo pulmonar.

O ecocardiograma transtorácico (ETT) mostrou uma dilatação marcada do ventrículo direito com septo interauricular intacto, sem hipertensão arterial pulmonar.

A ressonância magnética cardíaca (RMC) confirmou a dilatação ventricular direita e revelou hipocinesia marcada/acinesia da sua parede lateral. A prova de esforço foi negativa para isquemia.

À luz dos critérios de diagnóstico de displasia arritmogénica do ventrículo direito (DAVD), preconizados pela *Task Force* de 2010, este doente apresentava dois critérios *major* (na RMC: acinesia ou discinesia regional do VD e fração de ejeção $\leq$ 40%; no eletrocardiograma: ondas T invertidas nas derivações pré-cordiais direitas [V1-3]) e um *minor* (no ECGD-*Holter* de 24 horas > 500 extrassístoles ventriculares), pelo que o diagnóstico definitivo foi assumido.

Após o estudo eletrofisiológico, o doente foi submetido a implantação de cardioversor-desfibrilhador (CDI) monocâmara, encontrando-se assintomático desde então.

Esta apresentação tardia de DAVD evidencia a importância do rastreio por ETT, complementado por RMC. O risco de morte súbita cardíaca foi aferido pelo estudo eletrofisiológico, conduzindo à implantação do CDI. Estudos de associação genética devem ser oferecidos aos descendentes do paciente.

Case report

A 67-year-old man, independent in daily activities, a former pilot, with a history of essential hypertension and dyslipidemia (both under therapy), a current smoker (75 pack/years), and with no relevant family history, went to an emergency department due to an episode of general malaise accompanied by a sensation of constriction at the front of the neck and slight chest discomfort, followed by sudden loss of consciousness with complete and immediate spontaneous recovery. He reported no typical constricting chest pain, dyspnea, sweating, nausea or vomiting, asthenia, lower limb edema, focal neurologic signs, seizures or sphincter incontinence. There had been no further symptoms since that episode other than a feeling of general malaise.

Laboratory workup revealed negative troponin I and D-dimers. Arterial blood gases, posteroanterior chest X-ray, electrocardiogram (ECG), computed tomography (CT) angiography and brain CT showed no pathological alterations.

According to the patient, a provisional diagnosis of acute coronary syndrome was initially made and he was kept under observation for 24 hours. He was discharged the following morning without a definitive diagnosis.

The patient came to our emergency department 24 hours later with the same clinical setting except without loss of consciousness. He also reported a few seconds of palpitations. He denied other symptoms, and mentioned similar episodes a few weeks previously.

Physical examination was normal. The ECG showed sinus rhythm with no signs of acute ischemia, T-wave inversion in V1-V3 and absence of epsilon waves (Figure 1).

The posteroanterior chest X-ray showed no relevant alterations.

Blood tests revealed no abnormalities with no elevation of acute phase parameters, and renal function, electrolytes, coagulation tests, troponin I and CK-MB were within normal limits.

Transthoracic echocardiography (TTE) showed an undilated left ventricle with good global systolic function, no wall motion abnormalities and ejection fraction (EF) of 64%; massive right ventricular (RV) dilatation, with tricuspid annular plane systolic excursion (TAPSE) of 19 mm, dilated right atrium, <50% inspiratory collapse of the inferior vena cava and pulmonary artery systolic pressure of 31.60 mmHg; and no other relevant findings.

During hospital stay the patient's symptoms disappeared. Laboratory tests showed NT-proBNP of 705 pg/ml but no other abnormalities.

Given the unexplained loss of consciousness together with the presence of RV dilatation with no apparent alterations in wall motion or ventricular function, 24-hour Holter ECG monitoring, exercise testing and cardiac magnetic resonance imaging (MRI) were requested.

Holter monitoring revealed a mean of 22 isolated bimorphic ventricular extrasystoles per hour (528 in 24 hours), and six pairs. Exercise testing using the Bruce protocol had a duration of 7 min 30 s and was terminated by patient fatigue, at which point he had reached 90% of age-predicted