

When mitochondrial disease mimics a fulminant myocarditis: unusual onset of carnitine palmitoyl transferase II

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INTRODUCTION: We report the case of a 16-year-old patient, admitted at Bambino Gesù Hospital for cardiogenic shock. He was a competitive athlete with history of sporadic episodes of myalgias and elevated CPK. One week before he had an episode of headache and prolonged fasting during which he performed intense workout. He presented to ED for asthenia, chest pain and hypotension. The electrocardiogram showed sinus rhythm, normal PR and QTc intervals and low QRS complexes voltages in precordial leads. The echocardiography detected severe left ventricle (LV) dysfunction (EFbp 15%), mild hypertrophy and diameters dilatation with normal valves function and absence of pericardial effusion. Chest x-ray revealed mild congestion.

For the hemodynamic instability, the patient was first intubated and promptly treated for ventricular fibrillation, then, ECMO and Impella were inserted. Blood exams revealed increase of hs-I-troponin (2111 pg/ml), NTproBNP (20343 pg/ml) and CPK (> 22000 U/L). Rhabdomyolysis with acute kidney insufficiency has led to start veno-venous hemofiltration.

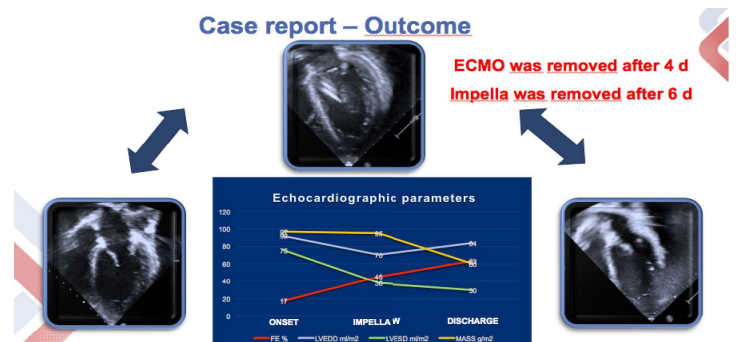
The endomyocardial biopsy showed no infiltrates with normal myocytes and edema. Toxicologic screening and metabolic test were performed. No abuse substances have been detected while biomarkers for metabolic disease confirmed suspicion for beta oxidation disease. LV recovered after 72 h and ECMO and Impella weaned easily.

Genetic study confirmed carnitine-palmitoyl transferase 2 deficiency (homozygous mutation and clinically useful variant c.77602C>T in heterozygosity of TTN gene), rare condition considered in beta oxidation disease.

After 1-month, cardiac magnetic resonance confirmed LV systolic function normalization and improvement of hypertrophy. The weighted sequences for tissue characterization showed non-specific significance changes. Cardiopulmonary test showed VO₂ of 40 ml/kg/min and an increase of lactates (8 mmol/l) after the peak. Nutritional advices and behaviours were provided to the family: a low-fat diet supplemented with MCT oil and rich in carbohydrates at main meals and snacks and avoiding prolonged fasting

Conclusion: New systems to treat cardiogenic shock allow to improve survival in patients with rare disease, unmasking conditions with clinical overlap. Mitochondrial diseases are usually poorly explored by cardiologist. The late onset of some of these group of disease, typically in the second-third life decade, should be taken in account in differential diagnosis with myocarditis.

Case report – Outcome



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