CLINICAL VIGNETTE

Rare indication for cardioverter-defibrillator implantation: propionic acidemia complicated by dilated cardiomyopathy and prolonged QT interval

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Propionic acidemia (PA) is a rare (1 in 100 000–150 000 persons) inborn error of the metabolism classified as an organic acidemia, inherited in an autosomal recessive pattern and caused by a deficiency of propionyl-coenzyme A carboxylase (PCC). Inhibited or reduced activity of PCC results in the accumulation of propionyl-coenzyme A and its metabolites: methylcitrate, 3-hydroxypropionate, triglyglycine, and propionyl glycine, leading to hypoglycemia, hyperammonemia, and hyperglycinemia.[1] Clinical presentation of PA includes life-threatening episodes of metabolic decompensation, but also neurological, hematological, gastroenterological, and cardiac manifestations with cardiomyopathies and arrhythmias.[2] Treatment of PA includes metabolic compensation, diet modification, carnitine supplementation, and enteral antibiotic therapy to reduce the burden of propionic acid–producing gut bacteria.[1]

We report a case of a 20-year-old man diagnosed with PA at the age of 7 days. Since the age of 16 years, he had experienced asymptomatic prolonged corrected QT (QTc, 490–504 ms), infrequent ventricular extrasystoles, and episodes of junctional rhythm lasting a few seconds (FIGURE 1). At the age of 20 years, he was diagnosed with dilated cardiomyopathy (DCM) with left ventricular ejection fraction of 40%. Since early infancy, the patient was treated by diet modification. To date, the course of the disease has been mild. The patient's intellectual development is normal.

At the time of DCM diagnosis, the patient was metabolically stable and did not require diet modification. Because of the predisposition to hypotension, only angiotensin-converting enzyme inhibitor was administered (ramipril, 1 × 1.25 mg/d). A month later, transthoracic echocardiography revealed left ventricular ejection fraction of 35%, but the patient remained asymptomatic.

Although the patient did not meet standard criteria for implantation of an implantable cardioverter-defibrillator (ICD), we considered information provided in case reports on sudden cardiac death in patients with PA and prolonged QTc, and decided to implant the ICD as a primary prevention of sudden cardiac death. Because of the progressing cardiomyopathy and reports describing the reversal of cardiac abnormalities after liver transplantation in patients with PA,[3] the case was evaluated by a multidisciplinary team and the patient was referred for liver transplantation.

Cardiomyopathy (mainly DCM) affects 9% to 23% of patients with PA, and prolonged QTc interval is observed in 70%. These conditions are associated with increased mortality and develop regardless of the metabolic control of the disease.[1] The pathomechanism of cardiac involvement in PA remains unclear. It is probably caused

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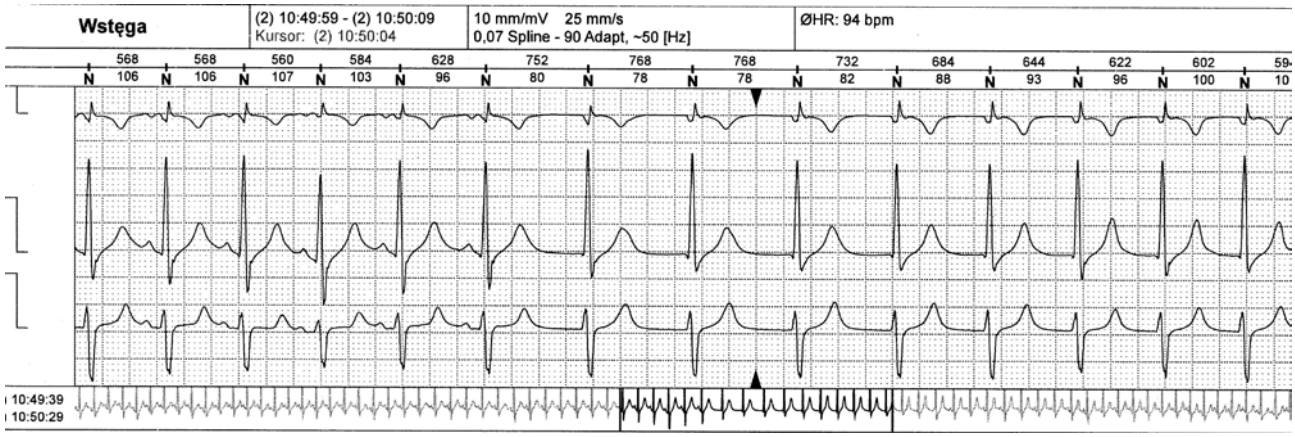

FIGURE 1 A 24-hour Holter recording: corrected QT interval prolonged to 490 ms in stimulations of the sinus node and an episode of nodal rhythm

by the altered energetic metabolism in cardiomyocytes. Other potential mechanisms include deficiency of carnitine and/or coenzyme Q10.[4,5] Low concentrations of carnitine were found in the heart in patients with PA, despite its sufficient oral supplementation and normal plasma concentrations.[2,5] Our study also revealed normal plasma carnitine levels.

The proposed guidelines recommend baseline and annual electrocardiography and transthoracic echocardiography, starting from the age of 6 years. This allows for a timely implementationof prophylaxis (eg, ICD therapy and avoidance of medications prolonging the QTc interval). Patients with cardiomyopathy or prolonged QTc interval should receive standard cardiac therapy.[1] Liver transplantation is another therapeutic option, since it may stabilize metabolism and reverse cardiomyopathy.[3]

ARTICLE INFORMATION

CONFLICT OF INTEREST None declared.

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