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Barth Syndrome and Left-Ventricular Non-Compaction: Case Report

Ece I. (1), Ture M.(1), Koca S.(2)
Yuzuncu Yil University Hospital, Van, Turkey (1) Yuksek Ihtisas Heart-Education and Research Hospital, Ankara, Turkey (2)

Introduction: Barth syndrome (BTHS) is an X-linked recessive disorder characterized by cardiomyopathy, skeletal myopathy and cyclic neutropenia in male patients. It is caused by mutations in the TAZ gene coding for the tafazzin, a protein involved in the remodeling of cardiolipin. We report a case of BTHS confirmed by TAZ gene analysis.

Methods:Three years old boy presented recurrent episodes of respiratory distress, progressive muscle weakness, hypotonia and growth failure. Family history was remarkable for 2 male siblings who died because of undetermined etiology.

Results:Hematologic and metabolic studies revealed that the patient had neutropenia, lactic acidemia, and 3-methylglutaconic aciduria; the diagnosis of BTHS was suspected. Echocardiography revealed mild dilated cardiomyopathy, hypertrophied and spongiform trabeculated pattern of the left ventricular posterior wall (ejection fraction= 58%, shortening fraction=28% (Figure 1). Electrocardiogram showed sinus rhythm with QTc duration of 460 ms, so propranolol 2 mg / kg was started and implantable cardioverter defibrillator fitted. Arrhythmia and pause not observed in the 24-hours rhythm Holter monitoring. The patient remained well at follow up 6 months later.
Conclusion: This case highlights that the diagnosis of BTHS should also be suspected in patients with left ventricular non compaction cardiomyopathy.