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Author`s Reply

To the Editor,

We would like to thank you for your criticism in this issue to our paper published in the Anatol J Cardiol (1). We appreciate the comments and want to briefly address the main questions raised in your letter. Noncompaction cardiomyopathy (NC) represents heterogeneity in its genetic pattern, pathophysiologic findings, and clinical presentations (2). The American Heart Association classified this entity as a primary genetic cardiomyopathy (3). According to the World Health Organization and European Society of Cardiology classification of cardiomyopathies, NC is still an unclassified cardiomyopathy (3-5). Additionally, there are several reports stating NC as genetic disorder and explain its inheritance and genetic cause (5). Because the laboratory investigations revealed hypergonadotropic hypogonadism and a pelvic MRI demonstrated the absence of overs, uterus, or prostate in our patient, we performed conventional cytogenetic analysis to identify whether any chromosomal abnormalities may be associated with these extra cardiac manifestations. Cytogenetic analysis demonstrated a 46, XX karyotype without any chromosomal abnormalities. We did not perform other techniques to investigate complex chromosomal re-arrangements and micro-aberrations. Techniques, such as FISH, CGH, and microarray, may identify the likely genetic etiologies. After evaluation of all the cardiac and extracardiac manifestations, dysmorphologic signs, and pedigree analysis, we investigated the most probable candidate gene LMNA mutations associated with cardiomyopathies. Direct sequencing did not reveal any mutations in the coding region of the LMNA gene. To identify the genetic cause of NC in our patient, other known genes associated with NC should be investigated.

The patient had generalized muscle wasting since the first hospitalization. It was most probably associated with heart failure. We referred the patient to neurology during the first admission, and cerebral MR was performed; however, it did not reveal cerebral atrophy, calcification, demyelination, or hydrocephalus. There was suspicion for microangiopathic vascular involvement. Nerve conduction studies or needle electromyography was not performed. Due to non-adherence to the medical treatment, there were recurrent hospitalizations with heart failure decompensation; however, ischemic stroke, seizures, or syncope was not observed. According to cerebral MR findings, there was no sign suggesting previous stroke(s). Because of mental retardation and non-adherence to medical therapy, oral anticoagulant therapy was not administered.

To exclude any arrhythmia, we monitored the patient with telemetry during hospitalization and performed 24-h rhythm Holter but did not detect any arrhythmia. The patient had three healthy brothers and a sister, two sisters and a brother had suddenly died in childhood from unknown reasons; however, an autopsy was not conducted. A brother and sister of the patient were examined by echocardiography; however, there were no abnormality. Rest of the family members were considered as normal.

In conclusion we did not detect any finding, suggesting neuromuscular disease in our evaluation. Coexistence of biventricular NC, genital and skeletal anomalies, and mental retardation led us to consider the presence of a syndrome.

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Thrombus formation during septal puncture

To the Editor,

We deeply appreciate Bilge et al. (1) for this study published in September 2014 issue of The Anatolian Journal of Cardiology entitled "Left atrial spontaneous echo contrast and thrombus formation at septal puncture during percutaneous mitral valve repair with the MitraClip system of severe mitral regurgitation: a report of two cases." It was reported in both cases that activated clotting time (ACT) of patients were higher than 250 s; however, it was not emphasized whether unfractionated heparin (UFH) was administered before or after septostomy. This issue is important in patients, particularly with atrial fibrillation (AF) due to risk of thrombus formation. We have reported a case of mitral stenosis and AF who was administered UFH after septostomy and developed thrombus right after trauma of puncture of interatrial