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Case Report Open Access

Duchenne Muscular Dystrophy

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Abstract

Duchenne Muscular Dystrophy (DMD) is an X-linked inherited neuromuscular disorder due to mutations in the dystrophin gene. It is characterized by progressive muscle weakness and wasting due to the absence of dystrophin protein that causes degeneration of skeletal and cardiac muscle. We present a case of Duchenne muscular dystrophy induced myocardial damage manifesting as acute myocardial infarctionina 12-year-old boy.

Keywords: Acute myocardial infarction; Duchenne Muscular Dystrophy.

Case Presentation

A 12-year-old boy presented to the emergency department with intermittent chest pain of 2 days' duration. Electrocardiography performed on arrival revealed anterior ST elevation (Figure1A).Cardiac troponin I concentration was 28.91ng per milliliter (reference range,0 to 0.04ng per milliliter).Echocardiographic examination found no abnormalities. Emergency coronary artery computed tomography angiography performed 30 min after presentation showed no stenosis of coronary artery (Figure 2).

Blood analysis in the boy revealed an elevated total creatine kinase level of 2236 U/Land creatine kinase-MB mass (CK-MB mass) level of 76.7 ng/ml. He presented with progressive proximal weakness of the lower limbs starting at 4 years of age followed by involvement of the upper limbs, which raised concern about Duchenne muscular dystrophy (DMD), it is characterized by progressive muscle weakness and wasting due to the absence of dystrophin protein that causes degeneration of skeletal and cardiac muscle. Family history revealed a similar disease in a second-degree cousin. Muscle biopsy from right quadriceps showed rounded small muscle fibers with evidence of degeneration and an absence of dystrophin protein. The molecular diagnostic of DMD involves a deletions/duplications analysis performed by Multiple Ligation Probe Assay MLPA. Thus we confirmed the diagnosis of DMD. His chest pain symptoms and ST-segment resolution (Figure 1B)were obviously improved by metoprolol, trimetazidine, coenzyme Q10, perindopril and other supportive therapies.

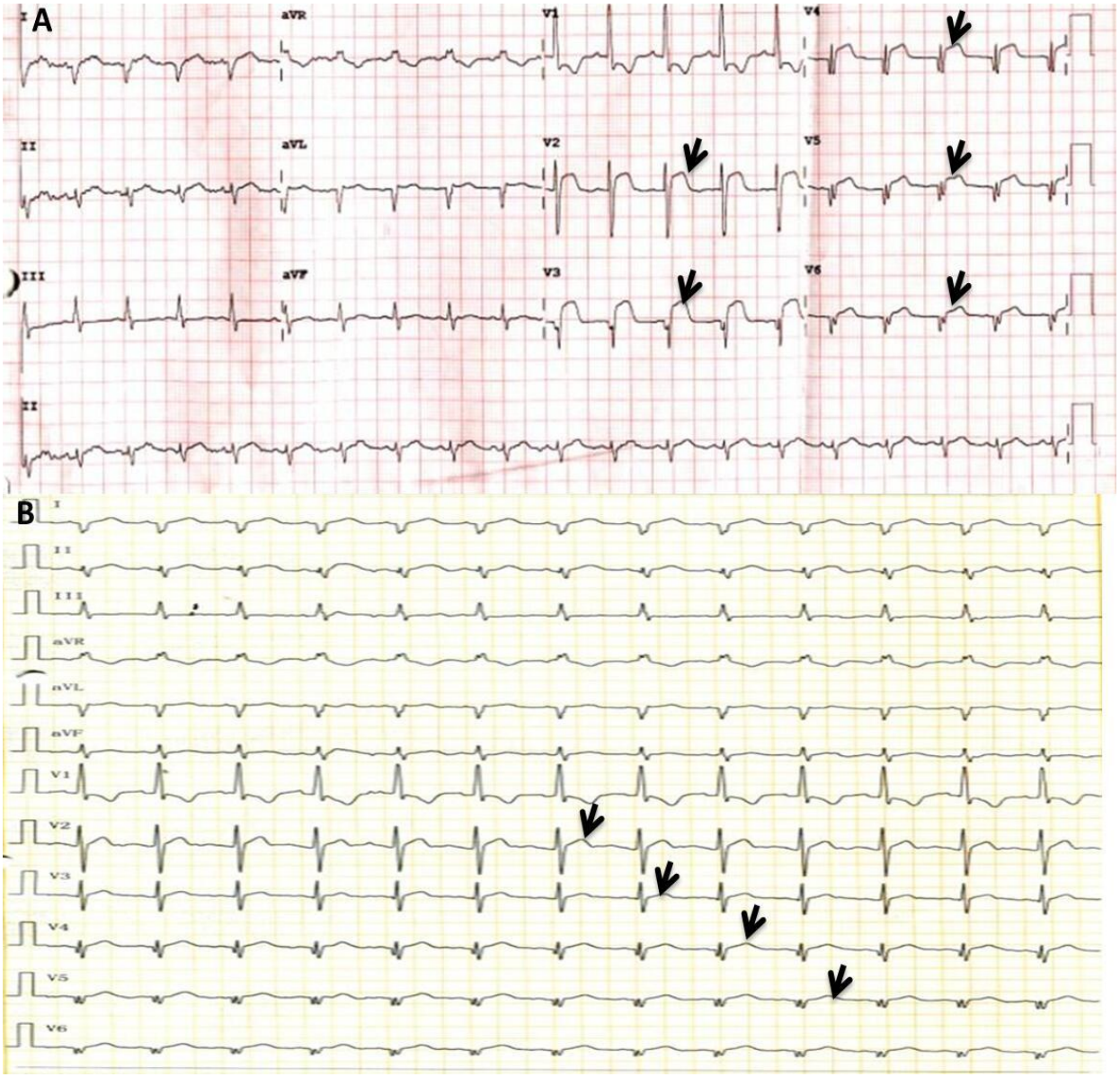

Figure 1: (A) On the electrocardiogram at emergency room, signs of acute myocardial infarction in the anterior territory were presented with ST elevation in leads V2 to V6 (**black arrow**). **(B)** On the electrocardiogram at discharge, signs of ST-segment dramatically resolution in leads V2 to V6 (**black arrow**).

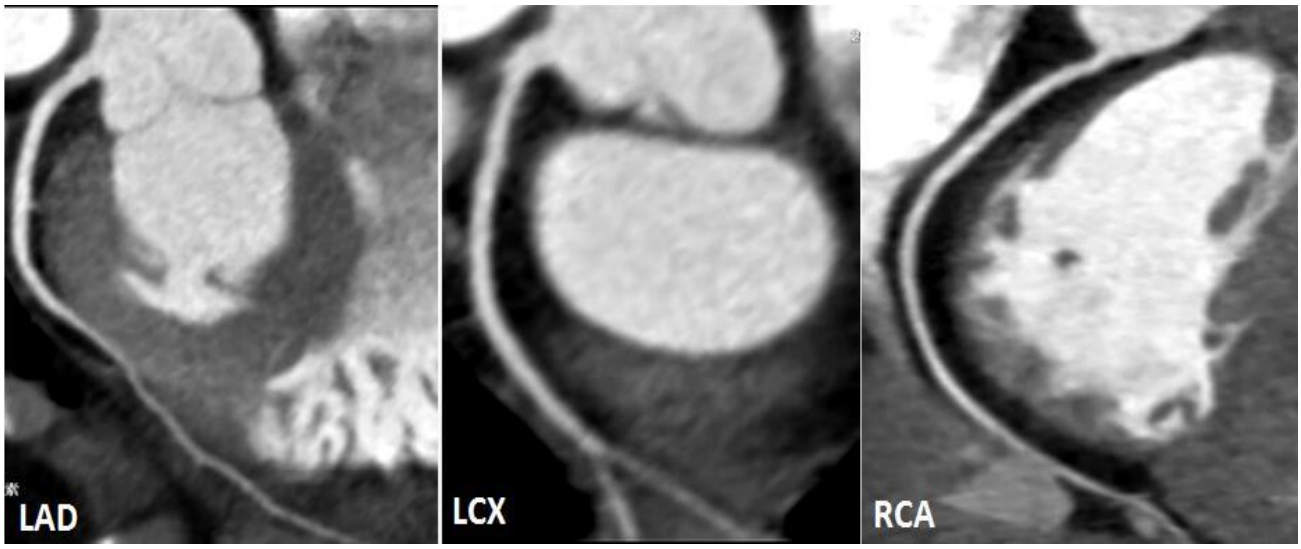

Figure 2: Emergency coronary artery computed tomography angiography performed 30 min after presentation showed no stenosis of coronary artery.

Discussion

DMD is an X-linked inherited neuromuscular disorder due to mutations in the dystrophin gene [1]. It is characterized by progressive muscle weakness and wasting due to the absence of dystrophin protein that causes degeneration of skeletal and cardiac muscle [2]. The molecular diagnostic of DMD involves a deletions/duplications analysis performed by quantitative technique such as microarray-based comparative genomic hybridization (array-CGH), Multiple Ligation Probe Assay MLPA [2]. DMD is the most prevalent neuromuscular disorders, affecting up to 1/3600 male births worldwide [3]. It is caused by mutations in the dystrophin gene on the X chromosome and the clinical signs are not present at birth [1]. The average age of diagnosis is usually at four years, when the first symptoms appear [4]. DMD patients develop a severe cardiomyopathy that generally manifests at about 10 years and is prevalent in most patients by 20 years of age [5]. Duchenne muscular dystrophy patients cardiac damage is mainly characterized by dilated cardiac myopathy, such as the left atrium, left ventricle enlargement, ventricular wall thinning, heart valvular regurgitation, reduced heart ejection fraction (EF) etc. [6, 7]. Electrocardiogram shows the narrow and deep Q waves, followed by sinus tachycardia, atrial flutter, PR interval period shortened [8, 9]. To date there have been 0 case report of DMD-induced STEMI. At present up to, beta receptor inhibitors and angiotensin receptor inhibitors had been showed can improve DMD-induced myocardial damage [10, 11]. His chest pain symptom, ST-segment resolution and myocardial injure was gradually improved by metoprolol and perindopril. Therefore, beta receptor inhibitors, angiotensin receptor inhibitors and increase myocardial metabolic could improve DMD-induced myocardial damage.

This case report is unique because it is first case report of DMD-induced STEMI. The learning points from the case report: firstly, DMD is a progressive inherited myopathy with an early onset in childhood [12]; secondly, it progresses to the bed-bound state in the second decade of life and patients usually succumb to cardiac complications; Again, beta receptor inhibitors, angiotensin receptor inhibitors and increase myocardial metabolic could improve DMD-induced myocardial damage [13]; Eventually, conservative management, active physiotherapy, genetic counseling and other supportive therapies hold the key to successful management of these cases.

To our knowledge, this is the first time that DMD induced myocardial damage manifesting as acute myocardial infarction. This discovery provides direct evidence for a myocardial damage, and lays out a foundation for DMD-induced myocardial damage intervention.

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