

A Huge Coronary Artery Aneurysm in Osteogenesis Imperfecta: A Case Report

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Abstract- Osteogenesis imperfecta (OI) as an inherited connective tissue disorder can affect all tissues that contains type I collagen. Well-known cardiac complications of this disease such as aortic root dilatation, aortic regurgitation and mitral valve prolapse have been rarely reported in the literature. Coronary artery aneurysm is a rare cardiac complication in OI, as reported in a 19 year old female presenting with myocardial infarction and hypotension.

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Introduction

Osteogenesis imperfecta (OI) is a rare, congenital, heredity, autosomal dominant disease. It is frequently caused by defect in the gene that produces collagen pro- α 1 and pro- α 2 chains, the most important protein block of the bone, which results in increased bone fragility and low bone mass. However, early and frequent clinical manifestations of this disease includes Brittle and deformed bones, dental abnormalities, short stature, hyper-extensible ligaments, blue sclera, early hearing loss, poor muscle tone and respiratory failure in severe cases. Other presentations such as cerebral (basilar) and cardiac abnormalities are infrequently reported due to presence of collagen in vessels (1,2). Several cardiovascular complications such as hypotension, larger left ventricular mass and diameter, mitral valve regurgitation and prolapsed valve, Aortic valve stenosis and regurgitation, tetralogy of Fallot (TOF), patent foramen oval (PFO), ascending, descending and abdominal aorta and aortic root dissection and other arteries dissection have been reported as case reports or low sample sized researches (3). Nevertheless – to the best of our knowledge- there are few reports of isolated coronary artery aneurysm (4). We are reporting a 19 year old female with type-I OI presenting with inferior

wall myocardial infarction and hypotension, which had multiple and huge aneurysms in her coronary arteries.

Case Report

Our case was a 19-year old bedridden female patient, known case of Osteogenesis Imperfecta from 4th year of her life. She had multiple hospital admission due to respiratory failure and was under treatment with continuous positive airway pressure (CPAP). In last admission, she was referred with shortness of breath, chest pain and hypotension (systolic blood pressure of 40 mmHg). An immediate ECG revealed inferior wall myocardial infarction (MI) (ST segment elevation in lead II, III and aVF) and a laboratory test showed an elevated cardiac markers (CPK= 338 mg/dl, CK-MB =77 mg/dl and troponin I= 0.793 ng/ml). She was admitted to the CCU for treatment and further investigation. After stabilization, an echocardiography was done which showed a large size (6 x 8 cm) non-homogenous mass adjacent to right atrium (RA) and right ventricle (RV), which compressed these chambers with suspicious of pericardial cyst. She was treated with diagnosis of MI. After discharge, she was referred for trans-esophageal echocardiography (TEE).

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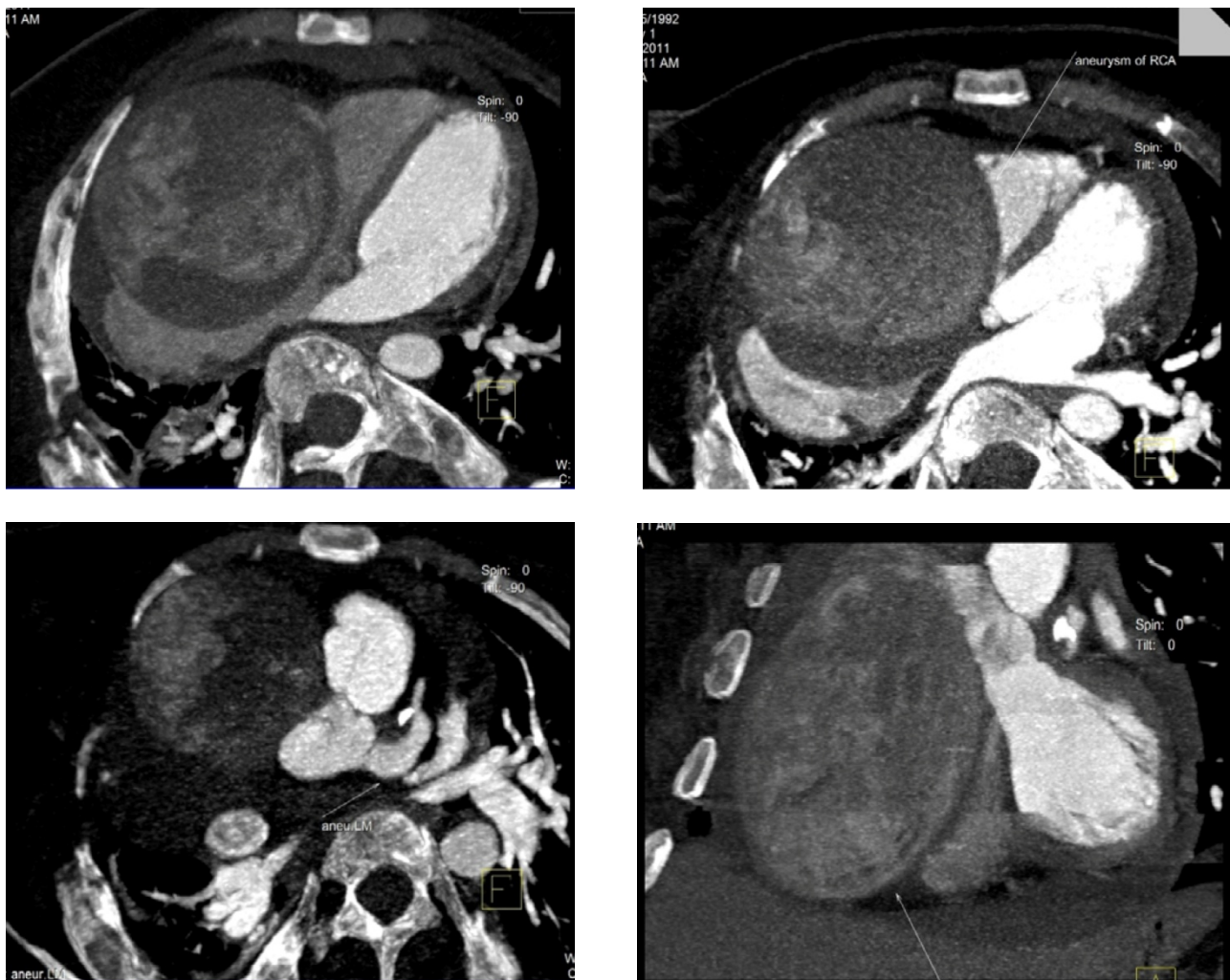


Figure 1. Huge aneurysm of right coronary artery (RCA) at the right side of the heart with compressive effect on right atrium and ventricle along with pericardial effusion. Mural thrombosis in the aneurysm is seen. Abnormal bony chest wall appearance due to known Osteogenesis imperfecta. A and B: MIP axial view; C: aneurismal dilatation of LM, LAD and Lcx; D: MIP coronal view

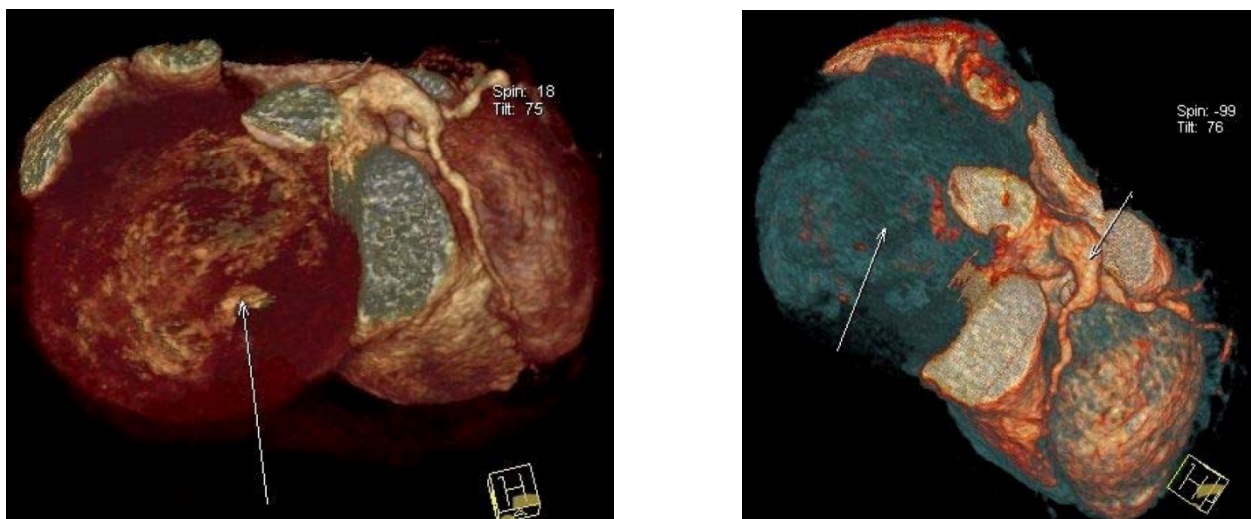


Figure 2. Volume rendering technique images. Huge right coronary artery aneurysmal dilatation (100 x 68 mm), which was compressing the aortic root and displacing right atrium and ventricle; the arrows indicate a huge right coronary artery (RCA) and a small Lcx aneurysmal dilatation

In TEE, LVEF was 55% with very large size (6 x 6cm) well-rounded non-homogenous cyst with Smokey appearance inside, which was compressing RA and RV. Due to history of MI, coronary CT angiography was requested and as seen in figures, 3D volume rendering images demonstrated a huge right coronary artery aneurysmal dilatation (100 x 68 mm), which was compressing the aortic root, displacing right atrium and ventricle. In addition, an 8 mm left circumflex artery aneurysmal dilatation accompanied with mild stenosis was seen. Due to its large size and her underlying disease, the medical committee board of the hospital decided to manage her conservatively without surgical procedures and follow her with serial echocardiography and laboratory tests. She was discharged with aspirin and clopidogrel with beta-blocker treatment.

Discussion

OI is an inherited disorder of type I collagen, which is classified into four major and four minor types. Most cases of OI are inherited from a parent, while some cases are the result of new genetic mutations. Type I collagen is mostly found in the bones; which OI presents with low energy trauma induced fractures. Type I collagen is also present in ligaments, sclera, teeth, skin, and other connective tissues; thus, patients with OI may have other related symptoms. However, other tissues without collagen type I involvement have been reported. (1,2).

As we know the endothelial monolayer of the vessels lies on a basement membrane that is made up of non-fibrillar collagen types, such as type IV collagen, laminin, fibronectin, and other extracellular matrix molecules (5). By aging, fibrillar collagen types such as type I and III adds to this combination; and type I collagen is affected in OI (6). Considering these substitutions of collagen by aging we can presumed that OI cardiovascular defects might become perceptible during the later years of patients lives. However, we cannot find a plausible reason why a young female OI subject can develop aneurysms.

There are over 20 cases reports on rare cardiac manifestations of OI, mostly followed by valve replacement in patients with valvular insufficiency or coronary artery bypass grafting (CABG) in patients with CAD (7). In one case report, there were three adults with OI reported with aortic aneurysms and valvular defects. The patients described in this report were 43, 47, and 50 years of age, two presented with aortic insufficiency due to dilatation of the aortic root and the third had lesions

involving pulmonary, aortic, and mitral valves in addition to microscopic changes in pulmonary artery and aorta (8). However, in a case report in 1987 from Houston, Texas, a 40 years man with OI was described with coronary aneurysm. The patient received saphenous vein aortocoronary bypass grafts to the left anterior descending, obtuse marginal and right coronary arteries with a 15-month follow-up negative for cardiac complication. The authors suggest CABG as a safe procedure in OI patents with gross atherosclerotic changes (4). However, our case did not have any risk factor for atherosclerotic changes and her coronary artery aneurysm was very huge. Considering the rarity of OI and the rarity of cardiovascular complication in this disorder and the low overall incidence of CAA, we can conclude that we are reporting a unique case of a huge (10 cm) RCA aneurysm in a young female with OI that has not been described in the literature until now.

Based on previous reports (4,7-8) coronary artery aneurysms were accepted as an independent predictor of mortality in patients with aneurysms. However, due to rarity and non-availability of controlled trials, there is no optimal management strategy for patients with giant coronary artery aneurysms. Thus, we hope such case reports could lead clinicians to establish an optimal protocol for management of coronary artery aneurysms and help us to recognize the unfamiliar aspects of cardiovascular complication in OI.

We believe that the coronary risk factors in patients with OI should be monitored and modified by clinicians. These monitoring should include regular cardiac and coronary function assessment in patients with OI.

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