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CASE REPORT

Brittle bones, fragile heart- a rare case report of osteogenesis imperfecta and mitral valve insufficiency with tragic outcome

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ABSTRACT

Osteogenesis Imperfecta is a rare disease of the connective tissue resulting in bone and tissue fragility. It is characterized by bone fragility, decreased bone mass, and frequent fractures. The range of phenotypic expression varies greatly, spanning from prenatal fractures resulting in fatal outcomes to milder presentations characterized by few fractures and typical stature. According to the Sillence classification that is based on clinical and radiological characteristics along with mode of inheritance it can be classified into 4 types. In this study, we report a 32 year old female with osteogenesis imperfecta who underwent mitral valve replacement. She was a known case of severe mitral valve incompetence secondary to Osteogenesis Imperfecta. She presented with shortness of breath, NYHA class 3 symptoms, and a history of unsuccessful previous percutaneous mitral valve repair. She underwent open mitral valve replacement with tissue valve owing to her risk of bleeding with anticoagulation. Unfortunately, despite meticulous surgical technique, the patient experienced post-operative complications that ultimately led to her demise.

Keywords: Osteogenesis Imperfecta; Mitral Valve; Mitral Valve Insufficiency; Hemorrhage; Blood Coagulation.

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INTRODUCTION

Osteogenesis imperfecta (OI) or brittle bone syndrome is a rare systemic hereditary connective tissue disorder with an incidence of about 1 in 10,000 to 20,000.[1] It's signs include fragile bones, (hence the name brittle bone disease) and multiple fractures.[2,3] It is mostly caused by mutations in the COL1A1 and COL1A2 genes, which encode for type I collagen.[4] Sillence et al classified OI into 4 clinical types: I, II, III, and IV; type I is mild disease, type II is lethal in the perinatal period, type III is a severe form, and type IV is of moderate severity.[5] In recent studies, it has been found that the morbidity and mortality rates of Osteogenesis Imperfecta (OI) are closely linked to abnormalities within the cardiovascular system. Cardiovascular abnormalities, such as aortic and mitral valvulopathies, atrial fibrillation, and heart failure, are frequently observed in patients with Osteogenesis Imperfecta (OI).[6,7] Collagen type 1 plays a significant role as a structural component in various regions of the cardiovascular system. These include the heart valves, chordae tendineae, fibrous rings of the heart, the interventricular septum, the aorta, and most arteries. Studies have reported aortic or mitral valve diseases as rare associations of OI.[8,9] Presented here is a case of a 32-year-old female patient with OI and severe mitral valve incompetence.

CASE PRESENTATION

A 32-year-old female from Afghanistan presented to us on May 10, 2023, following a MitraClip procedure done elsewhere, with chief complaints of persistent palpitations, shortness of breath, progressive dyspnea, New York Heart Association (NYHA) Class III functional class symptoms, that occasionally worsened to Class IV for the last few months. On admission, the patient exhibited vital signs of a heart rate of 110 beats per minute, blood pressure of 130/90 mmHg, respiratory rate of 20 breaths per minute, and an oxygen saturation of 92% on room air with BMI of 19.3kg/m^2. The cardiovascular examination revealed an irregular heart rhythm with a holosystolic murmur heard best at the apex, indicative of severe mitral regurgitation. Further examination demonstrated lateral scoliosis and a prominent rib cage cavity,

attributed to the underlying osteogenesis imperfecta. Her past medical history revealed multiple episodes of heart failure, with a recent diagnosis of severe mitral regurgitation. Additionally, she had a history of osteogenesis imperfecta (OI) with multiple bone fractures, and clinical examination revealed characteristic features of OI, such as short stature, blue sclera, sensorineural hearing loss, and prominent deformities of the limbs and skull. The past surgical intervention revealed the percutaneous intervention for mitral valve repair in the form of MitraClip, which failed to reduce the regurgitation or improve the symptoms. Comprehensive blood work, including a complete blood count and metabolic panel, revealed microcytic anemia with a hemoglobin level of 11.7 g/dL. Alkaline phosphatase (ALP) levels were elevated at 179 U/L, owing to multiple bone fractures. Imaging studies, including chest X-ray and CT chest was done to delineate the thoracic cavity anatomy which showed marked deformity of chest cavity & vertebral column (Figure 1). Echocardiography findings displayed severe mitral regurgitation, moderate tricuspid regurgitation, and anterior mitral leaflet prolapse into the left atrium with a posteriorly directed jet. Furthermore, a MitraClip was noted to be attached to the mitral valve apparatus, and a dilated left atrium with multiple small interatrial septal defects shunting left to right was also observed. Given the failure of the previous percutaneous mitral valve repair, the patient underwent evaluation for mitral valve replacement surgery as the definitive treatment for severe mitral regurgitation. Due to the complexity of her condition, a multidisciplinary approach involving cardiologists, cardiothoracic surgeons, and rheumatologist specialists in managing osteogenesis imperfecta-related complications was adopted. Before proceeding for surgery, we faced certain challenges such as choice between an open-heart and a percutaneous approach, and the selection of either a tissue or mechanical valve. Given the patient's history of failed MitraClip repair, the options for percutaneous intervention were limited.

Her mitral valve anatomy had become complex and challenging, making another attempt at a percutaneous approach less likely to succeed. Consequently, the team opted for an open mitral valve replacement to provide a more comprehensive and tailored approach. The open approach offered better visualization, access to the mitral valve, and the ability to address the anatomical complexities of the patient's case. Another crucial decision was whether to implant a tissue or mechanical valve. A mechanical valve would have required the patient to take lifelong anticoagulants to prevent thrombotic complications. Considering the patient's age, lifestyle, and her desire to avoid the potential complications and restrictions associated with anticoagulant therapy, we chose a tissue valve. This decision was made to optimize the patient's long-term quality of life and minimize the risk of bleeding complications. The patient was brought to the operating room, where general anesthesia was administered. Invasive lines were inserted, and sterile draping was meticulously performed. The surgery included a midline sternotomy, thymectomy, pericardiotomy, conventional distal ascending aortic, bicaval venous cannulation, and establishment of cardiopulmonary bypass (CPB). A cross clamp was applied to arrest the heart. Upon opening the left atrium at the Sonderguard groove, the mitral valve was meticulously examined. The previously implanted MitraClip (figure 1) was identified and subsequently explanted allowing examination of the mitral valve, and implantation of a 25mm porcine (tissue) St. Jude Medical (SJM) valve (Figure1) using pledgeted Ethibond suture. Following the successful valve implantation, the left atrium (LA) was closed after Dearing, and the cross clamp was removed. Weaning off CPB was achieved smoothly, and heparin was reversed with protamine. Post-bypass, the patient exhibited difficulty achieving hemostasis, despite transfusions of fresh frozen plasma (FFP) and platelets. Notably, her blood remained thin, and she experienced continuous oozing from multiple sites, leading to a drop in her hemoglobin levels. The initial arterial blood gases (ABGs) upon arrival in the intensive care unit (ICU) indicated a hemoglobin level of 4 g/dL. Suspecting internal bleeding due to persistent oozing, blood transfusions were initiated. Subsequent diagnostic imaging, including a chest X-ray (figure 1), showed no obvious collection in the pericardial or pleural cavity. However, the patient's condition deteriorated rapidly, with a significant drop in blood pressure, necessitating escalating inotropic support. Fresh blood was observed in the endotracheal tube (ETT), suggesting potential airway bleeding. Unexpectedly, the patient went into cardiac arrest. The chest was reopened at the bedside, and cardiac massage was performed for 2 minutes, successfully restoring sinus rhythm. However, no significant blood was found in the pericardial or pleural cavities during this intervention. Approximately half an hour later, the patient experienced another cardiac arrest. Despite 20 minutes of open cardiac massage, the patient could not be resuscitated and was declared deceased.

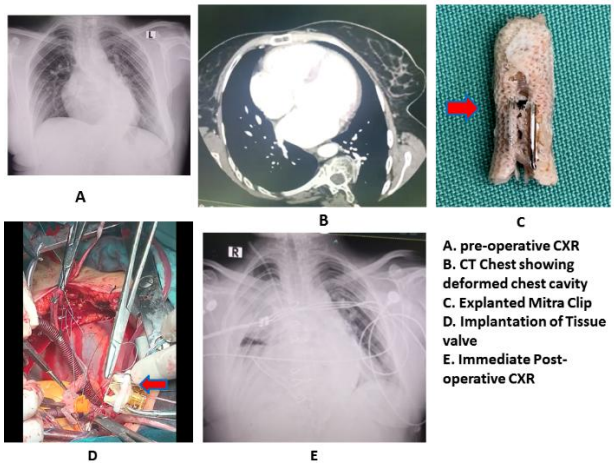

Figure 1: Pre-, Peri-, and Postoperative findings in the patient.

DISCUSSION

Osteogenesis Imperfecta (OI) is a hereditary connective tissue disorder with a population prevalence of 11 per 100,000 and is present in around 22 per 100,000 newborns.[8] According to reports, type I OI occurs in the population at a rate of 2.35 to 4.7 per 100,000 people worldwide. Type II OI is reported to occur between 1 in 40,000 and 1.4 in 100,000 live births. Types III and IV OI have a substantially lower incidence than type II, however its exact incidence is unknown.[9] OI manifests itself as blue sclera, middle ear and inner ear abnormalities, teeth, central nervous system, skin, and ligaments and tendons; however,

cardiovascular manifestations are rare.[10] Aortic root dilation and left sided valvular regurgitation are the most common reported cardiac pathologies in the literature,[11] like in our case presented above. Cardiac surgery has been related to higher morbidity and mortality in individuals with OI; 18% reported in one study.[11] The underlying connective tissue disorder that causes increased tissue and capillary fragility, platelet dysfunction, and impaired hemostasis which results in higher bleeding, poor and delayed wound healing which increases the risk of complications.[12] Our patient encountered the same complication, which could not be revived and ultimately resulted in her demise. This was attributed to the fragility of the tissues along with cardiac manifestations of OI.

CONCLUSION

Patients with collagen disorder may progress to uncontrolled coagulopathy after cardiopulmonary bypass and is major risk factor contributing to surgical decision making.

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