# Aortic valve replacement in a patient with ostegenesis imperfecta

## A case report

*Ann. Ital. Chir., 2014 85: 593-595*
pii: S0003469X14021253

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**Aortic valve replacement in a patient with osteogenesis imperfect. A case report**

AIM: *Osteogenesis imperfecta (OI) is an inherited connective tissue disorder in which fragile bones readily cause fractures. Aortic root dilatation, aortic valve regurgitation and mitral valve prolapse are uncommon cardiovascular manifestations of OI. Cardiac surgery in these patients carries a high risk of complications due to increased tissue and capillary fragility. We describe an open heart surgery in a woman with isolated aortic valve regurgitation secondary to OI.*
MATERIAL OF STUDY: A *58-year-old woman was referred to our hospital for surgical correction of aortic valve regurgitation. She had a past history of recurrent long bone fractures, and OI was diagnosed in the childhood. A standard median sternotomy was performed; the sternum was found to be thin and brittle. The native aortic valve was replaced with a size 23 mm stented aortic bioprosthesis. The sternum was closed with stainless steel wires.*
RESULTS: *The postoperative course was uneventful, and the patient was discharged home on the eighth postoperative day. We used thoracic band to avoid sternal diastasis. One year postoperatively, the echocardiogram showed a normal aortic bioprosthesis function without paravalvular leakage. The sternum was stable without dehiscence.*
DISCUSSION: *The mortality rate in cardiac surgery patients with heritable generalized connective tissue disorders, such as osteogenesis imperfecta, is high. Although tissue friability had no impact on surgical outcome, it should be kept in mind when operating on patients with OI.*
CONCLUSIONS: *We highlight the importance of a meticulous surgical technique, together with a strategy for management of anticipated perioperative complications to ensure a successful outcome.*

KEY WORDS: Aortic valve, Endocardirtis, Mitral valve, Replacement

## Introduction

Osteogenesis imperfecta (OI) is categorized in a group of heritable of connective tissue disorders. OI results from deletions, insertions or exon slice errors in the genes encoding type I collagen pro- 1 and pro- 2 chains. In most cases, the mutation is unknown and diagnosis is made by clinical assessments of symptoms. The clinical manifestations of the disorder can be skeletal (bone fragility, hyperextensible joints and ligaments), cutaneous (thin and translucent skin), ocular (keratoconus, megalocornea, blue sclerae), and dental (hypoplasia of dentin and pulp). Cardiovascular abnormalities are infrequently documented in OI. Aortic root dilatation, aortic valve regurgitation, and mitral valve prolapse are well known but are uncommon cardiovascular manifestations. Also rare are aortic dissections, significant coronary artery disease and coronary artery aneurysms. One case of spontaneous multivessel cervical artery dissection has been

*Pervenuto in Redazione Gennaio 2013. Accettato per la pubblicazione Marzo 2013*
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described [1], but no coronary artery dissection, either spontaneous or induced by catheter has been reported earlier in patients with osteogenesis imperfecta. Surgical procedures performed in these patients carry a higher risk of complications related to platelet dysfunction, tissue friability, impaired wound healing, and musculoskeletal weakness and deformity [2].
We describe a successful open heart surgery for aortic valve regurgitation in a woman with OI.

## Material and Methods

A 58-year-old woman was referred to our hospital for surgical correction of aortic valve regurgitation. She had a past history of recurrent long bone fractures, and OI was diagnosed in the childhood. On admission, the patient's height was 138 cm and weight was 53 kg, and blue sclerae were noted. Auscultation revealed a diastolic grade 4/6 murmur at the right sternal border and an Austin Flint murmur. Chest radiography showed moderate cardiomegaly. The electrocardiogram was unremarkable, except for voltage changes consistent with left ventricular hypertrophy. Transthoracic echocardiography showed a morphologically tricuspid aortic valve with evidence of severe aortic valve regurgitation. The left ventricle was dilated with an end-diastolic diameter of 62 mm with preserved left ventricular function. Aortic root diameter was normal. Tricuspid and mitral valve regurgitation were absent. Angiography demonstrated grade 4 aortic valve regurgitation with evidence of normal coronary arteries. Results of serum chemistry analysis, coagulation studies, and hematologic counts were normal.
A standard median sternotomy was performed; the sternum was found to be thin and brittle. Cardiopulmonary bypass was instituted with aortic and right atrial cannulation and the heart arrested with anterograde coronary ostial normothermic blood cardioplegia. The aortic wall was normal, and the valve was tricuspid and myxomatous. The native valve was replaced with a size 23 mm stented aortic bioprosthesis Edwards Perimount Magna (Edwards Lifesciences, Inc., Irvine, California) with interrupted 2-0 Ethibond supraannular sutures. The aortotomy was closed with a continuous 4-0 prolene suture and the patient was weaned from cardiopulmonary bypass uneventfully. We used Tabotamp® Fibrillar™ (Ethicon Inc, Sommerville, NJ, USA), a sponge-like collagen gauze patch of oxidized and regenerated cellulose that acts initially as a mechanical barrier for blood and subsequently becomes a viscous gelatine-like mass that serves as an artificial clot. The sternum was closed with stainless steel wires (Fig. 1).

Fig. 1.

## Results

No blood products were given during the operation. The patient was extubated six hours postoperatively.
The patient's postoperative course was uneventful and bleeding was limited to 650 mL in the first 24 hours requiring transfusion of two units of packed red cells. We used thoracic band to avoid sternal diastasis. Microscopic examination of the resected aortic valve showed myxoid degeneration without rheumatic degeneration.
One year postoperatively, the echocardiogram showed a normal aortic bioprosthesis function without paravalvular leakage. The sternum was stable without dehiscence.

## Discussion and Comments

OI is a heritable disorder of generalized connective tissues and the prevalence of OI is similar to that of Marfan's syndrome. Cardiovascular involvement in OI is rare compared with that of Marfan's syndrome [3]. Cardiovascular surgery has been reported in 41 patients with OI including our case, according to our survey of the English-language literature [4-9]. Aortic regurgitation (AR) is the most common valvular dysfunction in patients with OI, with an incidence of 1.8% [10], and is not related to either phenotype or severity of OI. Causes of AR in such cases include various dystrophic abnormalities of the aortic valvular apparatus such as thinning, sagging, and elongation of the cusps [10]. Dilatation of the annulus is common and bicuspid valves accounted for 30% of cases in one series [10].
The mortality rate in cardiac surgery patients with heritable generalized connective tissue disorders, such as Marfan's syndrome and osteogenesis imperfecta, is high. Six of the 41 OI patients who underwent cardiac surgery died postoperatively from hemorrhagic complications considered to be related to tissue friability [4-9]. A patient reported by Wong et al. who underwent valve replace-

ment required no perioperative blood transfusion, because the tissue was treated gently during surgery [5]. Closure of the sternum with sternal bands [7] and cardiac surgery via ministernotomy [9] to prevent chest trauma have been reported recently. Paravalvular leakage developing after double valve replacement has been reported[5]. We emphasize the importance of a planned surgical strategy and a meticulous surgical technique. The stress placed on the rib cage after careful sternotomy to avoid rib fractures can be decreased by minimal retraction. This should be followed by minimal and gentle tissue handling and dissection, and avoidance of unnecessary incisions on the aorta and adventitia. According to patient, we implanted an aortic bioprosthesis considering the future programmed orthopedic interventions and the high hemorrhagic risk of oral anticoagulant therapy in such patients.

Our patient's blood loss was only 650 mL in the following 24 hours and the chest drains were removed the following day. Gentle physiotherapy was instituted to ensure adequate lung expansion, but cautious enough to minimize chest wall trauma. He was discharged on the eigth postoperative day.

## Conclusions

This case report illustrates that mortality after cardiac surgery in OI patient may decrease if surgical treatment is done with care to details and tissue friability is kept in mind.


### Riassunto

L'Osteogenesi Imperfecta (OI) è un'alterazione ereditaria del tessuto connettivo in cui l'estrema fragilità ossea causa fratture. La dilatazione aneurismatica della radice aortica, l'insufficienza valvolare aortica ed il prolasso mitralico sono rare manifestazioni cardiovascolari di OI. L'intervento cardiochirurgico in questi pazienti può presentare importanti complicanze legate alla incrementata fragilità tissutale e capillare. Noi riportiamo un caso di chirurgia a cuore aperto in una donna con insufficienza valvolare aortica isolata secondaria ad OI. Il decorso postoperatorio è stato privo di complicanze maggiori e la paziente è stata dimessa presso propria abitazione in ottava giornata postoperatoria. È stata seguita con controlli frequenti per un anno. Nonostante la fragilità tissutale non abbia avuto impatto sul decorso postoperatorio, bisogna sempre tenere in considerazione la possibilità di eventi avversi nei pazienti con tale disordine connettivale. È necessario pertanto una meticolosa tecnica chirurgica che possa anticipare possibili complicanze postoperatorie.


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