A rare case report of coronary artery revascularization in a 16-year-old with familial hypercholesterolemia

Samir G. Marghade1*, Puneet Jandial2

1Department of Cardiothoracic surgery, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences, Sawangi (Meghe), Wardha, Maharashtra, India, 2Department of Cardiothoracic Surgery, Jabalpur Hospital and Research Center, Jabalpur, Madhya Pradesh, India

*Correspondence Author: Dr. Samir G. Marghade, Department of Cardiothoracic surgery, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences, Sawangi (Meghe), Wardha, Maharashtra, India.
Phone: +91-8408844455   Tel.: 00 91 7152 287702   Fax: +91-7152287714
e-mail: samirmarghade@gmail.com

Key words: Coronary artery bypass; Familial homozygous hypercholesterolemia
Received: 06.03.2015   Accepted: May 06.05.2015   e-published: 05.02.2016

Abstract

Familial homozygous hypercholesterolemia manifests as premature coronary occlusive heart disease. It is a rare dominant inherited disorder caused by the mutation at the locus encoding the low-density lipoprotein (LDL) receptor. Apart from coronary revascularization procedure, intensive cholesterol lowering therapy and LDL apheresis is needed. Very few cases have been reported in the literature so far. The subject of this case is a 16-year-old boy with a medical history of familial hypercholesterolemia who underwent triple coronary artery bypass grafting uneventfully.

Introduction

Familial homozygous hypercholesterolemia (FHH) is an autosomal dominant disorder caused by the mutation of the gene encoding the low-density lipoprotein (LDL) receptor [1]. Various manifestations of this disease include tendon xanthomas and cardiac involvement like premature coronary artery disease, valvular and supravalvular aortic stenosis in the natural course of the disease [2]. Being a rare condition, very few cases have been reported in the literature [3-8]. The surgical treatment of this condition is accompanied by a high degree of risk even in skillful hands [7]. Here, we report a 16-year-old boy with FHH, who underwent uneventful triple coronary artery bypass grafting (CABG) operation.

Case Report

We report the case of a 16-year-old boy who presented with intermittent retrosternal chest pain and dyspnea (NYHA II). He had a strong family history with his father who died due to myocardial infarction. His elder brother was dyslipidemic but was asymptomatic. He was detected to have FHH Type II b after genetic work up. Multiple tendon xanthomas were present on the extensor aspects of hand, elbow, knee, and ankle (Figure 1). Xanthelasma was also present on the medial aspect of eyes. Histopathology of these lesions showed foamy macrophages. He had grossly deranged lipid profile with total cholesterol 405 mg/dl, triglycerides 145 mg/dl, high-density lipoprotein (HDL)= 74 mg/dl, LDL 302 mg/dl, which was treated with aggressive lipid lowering therapy.

Electrocardiogram showed reversible ST depression in lateral leads. Coronary angiography showed significant lesions such as severe left main disease (70%), proximal right coronary narrowing (80%), and OM1 osteo-proximal (90%) with left subclavian artery stenosis. Color Doppler study
showed bilateral diffuse intimal thickening and multiple calcific plaques in both common and internal carotid artery with 50-60% narrowing in the internal carotid artery.

Apart from lipid lowering drugs, the patient was treated with aspirin, β-blockers, and sorbitrate. Off-pump CABG surgery was performed. Left internal mammary artery and two saphenous venous grafts were harvested. In view of left subclavian artery stenosis, proximal free end of the left internal mammary artery was anastomosed to the saphenous venous grafts to OM1. Both right coronary artery and OM1 received saphenous venous grafts. The surgery was uneventful, and the patient was discharged on post-operative day 7. The child was initiated on ezetimibe apart from Atorvastatin. However, the lipid control remained suboptimal with cholesterol levels more than 350 mg/dl. The patient could not undergo LDL apheresis and plasmapheresis due to cost constraints.

The patient remained symptom free on the 1st year follow-up.

Discussion

FHH is an inherited genetic disorder due to mutation of the gene for the LDL receptor (1). This condition is diagnosed by raised cholesterol and LDL levels in the absence of secondary hypercholesterolemia. Its incidence is 1:10,000,000 of the population [1]. Baker et al. stated a mortality of 100% at 30 years of age [2]. Very few cases have been reported in the literature and management is extremely challenging [3-8]. It manifests as premature coronary occlusive disease in second and third decade, however, earlier presentation is documented as well [3]. The other systemic involvement includes supravalvular and valvular aortic stenosis and tendon xanthomas [1,7]. In a study by Baghei et al., 11 out of 12 patients underwent primary CABG, and one underwent percutaneous coronary intervention. All patients received one internal thoracic artery and supplemental vein grafts [9].

Arterial grafts have been found to be superior in terms of actuarial survival and freedom from symptoms in these patients [10]. The use of arterial conduits for coronary problems in infants and children has been well-accepted in Kawasaki’s disease, post arterial switch and other congenital coronary lesions [11]. However, no additional benefits have been mentioned in using multiple arterial grafts [12]. The use of internal mammary arteries and gastroepiploic arteries has been well appraised in children [4]. Experience with radial artery has been limited in children [4].

Differences in adaptation to the growth of children have been shown between arterial and venous grafts. The thoracic artery grows in proportion to somatic growth whereas saphenous vein grafts tend to course in a more linear way with no increase in length or diameter [13,14]. However, isolated reports have highlighted excellent growth and patency of venous grafts up to 22 years after bypass surgery [13]. The patency of autologous saphenous grafts is 65.4 ± 7.9%, 84 months after operation in the patient more than 8 years of age [14].

Our patient had left subclavian artery stenosis hence free LIMA was grafted to the proximal SVG graft to OM1. Anastomosis between aorta and LIMA is technically complex because of the latter’s small caliber and the difference in thickness of both vessels. In such cases, the IMA can be anastomosed to the origin of a venous graft [15].

Intensive lipid lowering drug therapy reduced the frequency of progression of coronary lesions and increased the frequency of regression of coronary lesions. Thus, it reduced the incidence of cardiovascular events in men with coronary artery disease and high levels of LDL [16,17]. Lower the plasma cholesterol levels, the greater the likelihood that coronary artery disease can be prevented or delayed [18]. LDL apheresis is highly effective and selective in lowering LDL levels while leaving HDL levels unchanged. To conclude, in patients with FHH primary CABG especially IMA grafting increased the short-term freedom from reoperation and improves long-term survival [9,19].

Acknowledgments

We would like to thank the contributing author Dr. Puneet Jandial for guiding me throughout the process of manuscript preparation and final editing of the article. I am always indebted for his support. We would also like to thank my wife Dr. Pallavi Marghade for proof reading and editing the final manuscript.

References

A rare case report of coronary artery revascularization in a 16-year-old with familial hypercholesterolemia

Samir and Puneet


