Familial hypercholesterolemia (FH) is a genetic disease caused by a mutation in low-density lipoprotein (LDL) receptor gene. It causes various presentations including tendon xanthoma and cardiac manifestations. Herein, we present a young patient with homozygous FH (HFH) who presented with dyspnea and chest pain caused by coronary arteries stenosis and treated with coronary artery bypass graft (CABG) surgery at the age of 13 years. To the best of our knowledge, he is one of the youngest patients in the English language literature for whom coronary revascularization has been done in childhood.

Abstract

Familial hypercholesterolemia (FH) is a genetic disorder, which leads to premature coronary artery disease. The mutation in low-density lipoprotein (LDL) receptor gene causes various presentations including tendon xanthoma and cardiac manifestations. Such complications present in the early years of life in homozygous type of the disease [1, 2].

Coronary artery disease in childhood has been well documented in homozygous FH (HFH) [2–4]. During the natural course of the disease aortic valve involvement will develop that might restrict the patients’ activities and leads to progressive symptoms [5, 6].

Herein, we present a young patient with HFH who presented with dyspnea and chest pain caused by coronary arteries stenosis and treated with coronary artery bypass graft (CABG) surgery at the age of 13 years.

2. Case presentation

A 13-year-old boy was presented to the cardiologist with the chief complaint of dyspnea and chest pain after exertion. The history of the patient showed that he had documented evidence of hypercholesterolemia since four years of age, with total cholesterol level > 10.34 mmol/l. He had received lipid lowering drugs such as atorvastatin and cholestyramine. But continuous episodes of chest pressure and dyspnea were reported. Family history showed that the patient was from a consanguineous marriage and his father and his mother were cousins and both had hypercholesterolemia. His father had been operated on for three-vessel coronary artery disease two years before at the age of 39 years and both parents were taking anti-lipid medications. He had also positive family history for hypercholesterolemia in two of his cousins at the ages of 13 and 18 years. And his third cousin died of premature cardiac arrest at the age of 40 years.

In the last presentation, transthoracic echocardiography showed ejection fraction of 60%, supravalvular mild aortic stenosis (gradient = 10–12 mmHg), and mild mitral regurgitation. Because of the patient’s history, coronary angiography was done which showed left circulation dominancy accompanied by diffuse left main coronary artery lesion with 60% stenosis (Fig. 1) and 50% stenosis in the middle part of the left anterior descending (LAD) artery.

He was referred to us in March 2009. On admission to our ward, he had xanthelasma at the medial part of both his eyes and xanthoma on both hands, both elbows, popliteal area, and his buttock (Fig. 2). His total cholesterol level was 19.18 mmol/l, LDL was 12.25 mmol/l, and HDL was 5.15 mmol/l.

The patient was scheduled for CABG. After general anesthesia and via a midline incision the chest was opened and, by using the on-pump technique, we bypassed the LAD artery with left internal mammary artery (LIMA), and optuse marginal (OM) and left circumflex (LCX) with saphenous vein. LIMA and saphenous vein had good quality with thin wall without atheromatous plaques. Their diameters were 2 mm and 3 mm, respectively. The length of the LIMA graft was 15 cm and those used to bypass OM and LCX were 10 cm and 15 cm, respectively. The vessels were meticulously harvested using open classic method without trauma and they had good flow. The sutures were all continuous;
may present in childhood with premature coronary artery disease and xanthoma in various parts of the body. It is believed that total cholesterol level exceeding 9.68 mmol/l in children whose first degree relatives have hypercholesterolemia is highly suggestive for the disease [7].

Resulting atherosclerosis might lead to cardiac ischemia and coronary revascularization can be the last resort to alleviate the symptoms. Some researchers believe that arterial revascularization can be the best approach for the affected children [8], however, acceptable results from using venous grafts have been reported previously [2, 3, 6, 9]. We used LIMA to bypass the LAD, but as the patient’s coronary circulation was left dominant we could not use right internal mammary artery (RIMA) to bypass LCX and two venous grafts were used. Besides considering our previous experiment [6] and the good quality and size of the saphenous vein, and that the patient was scheduled for liver transplantation which could resolve the base of his disease, we were heartened to use such venous graft to bypass LCX and OM.

We performed a stress test one month later and found no signs of ischemia, so we did not perform the invasive angiography procedure.

Lipid lowering medications and lipid apheresis are two options to decrease serum cholesterol level; however, the best option to normalize the lipid profile might be liver transplantation [10]. Fortunately, a matched donor was identified within a three-month period after the CABB and the patient was transplanted successfully. Follow-up visits after discharging the patient from the transplantation ward showed low serum cholesterol and no complaint related to myocardial ischemia.

3. Discussion

The prevalence of FH has been reported differently worldwide and seems higher in our region [7]. Patients with FH

the distal anastomosis of the vein grafts was done using 7-0 Prolene and that of LIMA was done using 8-0 Prolene. The proximal anastomosis of the vein grafts was done by 6-0 Prolene.

The postoperative course in the ICU was uneventful and he was discharged home after three days. He was prescribed atorvastatin (40 mg/day) accompanied by diet modulation. The patient was referred as well to the liver transplantation unit. After one month we performed a stress test and found no ischemic changes. In a follow-up three months later, his total cholesterol was 19.39 mmol/l and his chest pain was resolved. In the last follow-up liver transplantation had been done, his serum cholesterol level was 3.23 mmol/l and he did not have any complaint related to cardiac ischemia.

References