An Uncommon Association of Sever Early Coronary Artery Disease and Aortic Coarctation in Patient with Familial Hypercholesterolemia: A Case Report

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Abstract

Familial hypercholesterolemia is a rare inherited disease that results in elevated levels of low-density lipoprotein (LDL) and clinically leads to early coronary artery disease. It is rare for this disease to be associated with the coarctation of the aorta. In this report, we review the clinical case of an 18-year-old who was referred to our center with angina pectoris manifestation. A comprehensive clinical and investigative evaluation of the patient pre-operatively showed the presence of familial hypercholesterolemia, Coarctation of the aorta, and ischemic cardiomyopathy that required coronary artery bypass surgery.

Introduction:

The familial hypercholesterolemia was thought to be caused by a genetic defect in the mechanism of cholesterol formation in the body, but current research confirms a defect in the catabolic of low-density lipoprotein (LDL)[1]. The LDL receptor function decreased due to a genetic defect; therefore, LDL receptor-mediated endocytosis is decreased leading to markedly elevated LDL levels[1]. This protein accumulates in the endothelium of blood vessels in the form of atherosclerotic plaque in the coronary arteries and the proximal aorta at an early age, leading to coronary artery disease and narrowing of the carotid artery. Arcus cornealis (figure1) is a single corneal circle of lipid deposits parallel to the limbus that usually affects both eyes symmetrically. Xanthomas (figure2) are yellow-colored deposition under the skin, characterized by accumulations of lipid-laden macrophages, can appear anywhere in the body in various disease states. Xanthelasma is a sharply demarcated yellowish deposit of cholesterol underneath the skin that occurs on or around the eyelids.

About 70-95% of cases of familial hypercholesterolemia (FH) are caused by heterozygous pathogenic variants. In contrast, homozygous FH patients have very high levels of LDL, an accelerated progression of atherosclerosis, and develop coronary artery disease at an early age.[2]

Case report:

A young patient, 18 years old, since he was 9 years old, was diagnosed with familial hypercholesterolemia of the homozygous type and has been receiving plasma apheresis periodically in our center since then. In preparation for liver transplantation, he received a cardiovascular system evaluation. During the evaluation, unexplained dyspnea was observed, and the patient was referred to the cardiac center to study the heart more broadly. Initially, the clinical cardiovascular evaluation showed the presence of a diastolic murmur in the aortic focus without a radio-femoral delay and the blood pressure measurements were within normal limits.

Imaging using echocardiography showed that the ejection fraction of the left ventricle is equivalent to 55% and the tri-leaflets aortic valve with mild to moderate aortic valve insufficiency. he had a cardiac CT

scan, which showed the presence of multiple coronary artery stenosis (figures3,4), as well as the presence of stenosis in the descending aorta after the branching of the left subclavian artery (figures5,6). A left cardiac catheterization was performed which showed stenosis of 20% in the left mean, with stenosis at the origin of the anterior descending artery by 80% and stenosis at the origin of the right coronary artery by 90%. The extensive study of the thoracic aorta revealed an acceleration in blood flow in the proximal part with a peak pressure gradient of 57 mm.

Carotid angiography showed stenosis of 70% in the right carotid artery with atherosclerosis in the left carotid artery without significant stenosis. The vascular surgeons and neurologists were involved, and given the necessity of coronary revascularization, they indicated that cardiac intervention is possible with a moderate risk of stroke in the post-operative period.

The case was discussed in a multidisciplinary meeting, where a consensus was made to perform coronary artery bypass grafting and to postpone the intervention on coarctation for a later stage after recovery from coronary injury and until the stenosis becomes more severe. In October 2020, the patient underwent complete arterial revascularization using the left internal thoracic artery and implanted it on the anterior descending artery, as well as implantation of the right internal thoracic artery on the right coronary artery. The patient spent a stable recovery period in the hospital for a week after the operation and was discharged without any complications.

Discussion:

The association between familial hypercholesterolemia (FH) and coarctation of the aorta is very rare. According to our best of knowledge, there is one case report describing this association.[3]

The presence of familial hypercholesterolemia requires special attention from the attending physician to the risk of developing coronary disease at an early age and without significant clinical symptoms.

Although the common age for the development of coronary disease in this type of patient is the end of the second decade and the third decade, many reports have indicated the possibility of developing coronary disease at an earlier age, especially in patients with poor control of the level of cholesterol or patients of the type homozygous.

The management of patients with FH with coronary disease depends on two parts, the first is medical and the second is surgical, as there is no long-term benefit from surgery unless total cholesterol level is strictly controlled, the researches indicate that achieving a total blood cholesterol level of less than 220 mg is of utmost importance to prevent the development of existing coronary disease as well as to prevent the occurrence of new coronary injuries.[4,5]

The benefits of using arterial grafts in particular in FH patients have been documented in several studies in terms of long-term survival as well as freedom from coronary revascularization.[6,7]

The comparison between the use of the right internal thoracic artery as a second graft (after using the left internal thoracic artery for grafting the anterior descending artery) and the saphenous vein is demonstrated by a meta-analysis of the superiority of the right internal thoracic artery in terms of long-term survival with a higher risk of deep sternal wound infection.[8]

The presence of coarctation of the aorta causes harm to coronary disease because it causes an increase in left ventricular end-diastolic pressure and thus resistance to blood flow in the coronary arteries.

Treatment of aortic coarctation is of great importance for coronary disease, but since the narrowing is not severe, its treatment can be postponed to a later stage.

Conclusion:

The association between coronary artery disease and coractation of the aorta in patients with familial hypercholesterolemia is rare. The physician should be careful of the hidden development of coronary artery

disease in these patients. Best management of these conditions requires a heart team meeting to determine the best intervention time and method for each patient.

IRB and informed consent statement :

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The data that support the findings of this study are available from the corresponding author, upon reasonable request.

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Figures Legends:

Figure1: Arcus cornealis in the left eye results from cholesterol infiltration around the corneal rim.

Figure2: Sever bilateral knee xanthoma.

Figure3: Curved multi-planar reconstruction of the right coronary artery showing non-calcified plaque causing significant ostial stenosis (yellow arrow).

Figure4: Curved multi-planar reconstruction of the left anterior descending coronary artery non-calcified plaque causing significant ostial stenosis (yellow arrow) and partially calcified plaque causing significant stenosis of the proximal segment (red arrow).

Figure5: 3D volume rendered image of the ascending aorta, arch and descending thoracic aorta showing significant coarctation (yellow arrow).

Figure6: Aortic geometry reconstruction of the aorta showing double oblique image of the coarctation site (A), luminal view (B) and curved multi-planar reconstruction (C).

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