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FAMILIAL HYPERCHOLESTROLEMIA WITH PREMATURE CORONARY ARTERY DISEASE AND CONCURRENT RENAL ARTERY STENOSIS

Cardiology

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ABSTRACT

Familial hypercholesterolemia (FH) is an inherited lipid disorder associated with markedly elevated low-density lipoprotein cholesterol (LDL-C) and premature coronary artery disease (CAD). We report a rare case of a young adult presenting with early-onset myocardial infarction and poorly controlled hypertension, later diagnosed with concurrent renal artery stenosis (RAS). Genetic testing confirmed heterozygous FH. This case highlights the aggressive atherosclerotic potential of FH beyond coronary vessels, emphasizing the importance of early diagnosis, comprehensive vascular assessment, and targeted lipid-lowering therapy. Multidisciplinary management, including revascularization and intensive statin therapy, is essential to prevent further cardiovascular and renovascular complications in such high-risk patients.

KEYWORDS

INTRODUCTION:

Familial hypercholesterolemia (FH) is an underdiagnosed hereditary disorder affecting LDL-C metabolism. It is strongly associated with atherosclerotic cardiovascular disease (ASCVD), increasing the risk of coronary artery disease (CAD) by up to 20-fold. FH is an autosomal dominant genetic disorder, and early identification is essential. High-intensity statins can reduce LDL-C levels by up to 50%, playing a crucial role in management.[7]

CASE:

We present a case of a 40-year-old female with a history of diabetes mellitus, poorly controlled hypertension (systolic BP: 180–200 mmHg), and non-ST elevation myocardial infarction (NSTEMI). She reported angina-like chest pain for six months. Coronary angiography revealed severe triple vessel disease: 90% occlusion of the left main coronary artery (LMCA), 80% in the left circumflex (LCX), and 95% occlusion of the right coronary artery (RCA). She underwent coronary artery bypass grafting (CABG).shown in figure 1 Despite treatment with three antihypertensives and statins, the patient had persistent hypercholesterolemia. Physical examination revealed xanthelasmas, corneal arcus, and tendon xanthomas over the elbows and Achilles tendons. Bilateral renal and carotid bruits were noted. Family history was significant for premature CAD in her brother and elevated cholesterol in her daughters.

Laboratory findings showed total cholesterol of 460 mg/dL, triglycerides of 276 mg/dL, LDL of 372.8 mg/dL, and an LDL/HDL ratio of 11.65. Multi-slice CT imaging confirmed diffuse atheromatous vascular changes and >60% bilateral renal artery stenosis, more severe on the left side.

This case underscores the importance of early diagnosis and comprehensive management in FH, including vascular screening beyond coronary arteries. Multidisciplinary intervention, including lipid-lowering therapy and revascularization, shown in figure 2 is vital to prevent progressive cardiovascular and renovascular complications.

DISCUSSION:

Familial hypercholesterolemia is a panvascular disease, and its complications are not limited to early-onset coronary artery disease but may extend to early-onset hypertensive renovascular disease and other atherosclerotic conditions, such as mesenteric vessel involvement, which should be distinguished from essential hypertension.[1]

Limited data report the role that certain renin–angiotensin system genetic polymorphisms can play in accelerating coronary heart disease in patients with familial hypercholesterolemia. Stenosis of the renal arteries[3], a well-known cause of secondary hypertension, is commonly due to atherosclerotic plaque, often found in the proximal to mid portion of the artery. Although a clear benefit of renal artery stenting over optimal medical therapy has not been demonstrated in non-FH individuals with atherosclerotic renal artery stenosis (RAS) [4], no focused studies have been conducted in FH patients.[2]who may be more likely to develop early-onset symptomatic RAS.

Treatment based solely on the degree of RAS may lead to further disease progression and ultimately renal atrophy[5]. The identification of RAS in patients with FH and hypertension may provide valuable information to further refine the recently developed SAFEHEART risk prediction equation for long-term ASCVD risk in FH.[6]

CONCLUSION:

Familial hypercholesterolemia is a systemic atherosclerotic disorder that often remains subclinical and must be proactively investigated. Patients presenting with premature atherosclerotic cardiovascular disease (male <55 years, female <60 years) and uncontrolled hypertension with renovascular pathology should be screened for familial hypercholesterolemia, as early and intensive pharmacological intervention can help prevent serious atherosclerotic complications

FIGURE LEGEND:

Figure1: Coronary Angiogram (rao) view

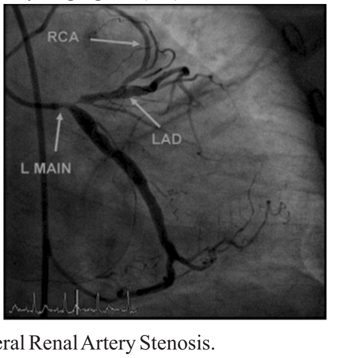
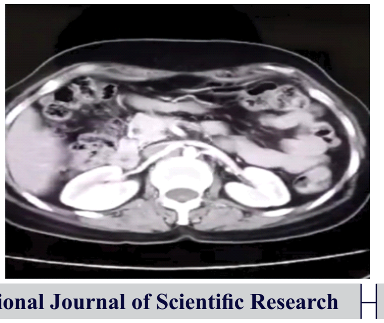


Figure2: Bilateral Renal Artery Stenosis.



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