A 34-year-old man was admitted to the hospital because of worsening of exercise tolerance and exertional dyspnea for about 3 months, without chest pain. A medical history revealed familial hypercholesterolemia (FH) diagnosed 8 years earlier (also in his brother), which was treated with rosvustatin (20 mg) and ezetimibe, to poor effect. On admission, an electrocardiogram showed sinus rhythm and left ventricular hypertrophy (FIGURE 1A). Laboratory tests showed the following lipid levels: total cholesterol, 8.19 mmol/l; low-density lipoprotein cholesterol, 5.9 mmol/l; high-density lipoprotein cholesterol, 1.76 mmol/l; and triglycerides, 1.19 mmol/l; moreover, the patient had elevated levels of liver enzymes (alanine transaminase, 72 U/l; aspartate transaminase, 63 U/l) and brain natriuretic peptide (527.7 pg/ml). Troponin levels were within the reference range.

Transthoracic echocardiography showed enlargement of the left atrium (48 mm) and the left and right ventricles (65 mm and 33 mm, respectively), with thin walls and restrictive flow through the mitral valve, with moderate mitral regurgitation and increased trabeculation of the apex and apical and mid segments of the lateral
Left ventricular noncompaction cardiomyopathy, severe CAD, and FH

Moreover, it revealed global hypokinesis with an ejection fraction of about 25%. Cardiac magnetic resonance confirmed the diagnosis of left ventricular noncompaction cardiomyopathy (noncompacted to compacted layers, 2.9; FIGURE 1E and 1F). Coronary angiography revealed diffused 3-vessel disease with critical stenoses of the left main coronary artery (LMCA), left anterior descending artery (LAD), intermediate branch and chronic total occlusion of the circumflex artery, and severe stenosis of the right coronary artery (RCA) (FIGURE 1G and 1H).
The patient was not referred for coronary artery bypass grafting because of a poor availability of the peripheral portions of the coronary arteries. A percutaneous coronary intervention (PCI) of the LMCA/LAD and intermediate branch with drug-eluting stent implantation (everolimus) was successfully performed (FIGURE 1I and 1J). A PCI of the RCA is the next scheduled procedure. If standard pharmacotherapy for severe hypercholesterolemia proves ineffective, a therapy with PCSK9 inhibitors will be considered. The patient is still alive.

Premature vascular disease with an associated high risk for cardiovascular events is common in patients with FH. Effective control of cholesterol levels is highlighted as one of the most important factors in patients with coronary artery disease (CAD). The identification of patients with true FH by evaluating the positive clinical sings of FH as well as by genetic testing might be useful for risk stratification for CAD among patients with severe hypercholesterolemia. However, the lack of significant differences in arterial stiffness between patients with and without FH may indicate that the FH mutation itself is not the main determinant of endothelial dysfunction and vascular remodeling in younger patients with hypercholesterolemia. The presence of severe CAD in a young patient with FH has been described before, but to our knowledge, this is the first report of the coexistence of CAD and FH with left ventricular noncompaction cardiomyopathy.

FIGURE 1 I–J – coronary angiography after successful PCI

REFERENCES


