A case of heterozygous familial hypercholesterolemia presenting with premature cardiovascular event

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Familial hypercholesterolemia (FH) is an autosomal dominant disorder that occurs due to mutations in the low-density lipoprotein (LDL) receptor gene located on chromosome 19. We report a case who was treated suboptimally for dyslipidemia for a long time and eventually diagnosed as FH after premature cardiovascular disease (CVD). A 45-year-old man with history of dyslipidemia and gout visited outpatient clinic for unstable angina. He had eyelid xanthomas (Fig 1.) and corneal arcus (Fig 2.). He also had familial history of hypercholesterolemia (mother, siblings). His electrocardiogram showed sinus rhythm and ST depression in inferior leads. Laboratory findings revealed mildly elevated high-sensitive troponin-I 21.3ng/L. His LDL cholesterol level was 281mg/dL and triglyceride level was 129mg/dL . Coronary angiography (CAG) was performed and showed 3 vessel disease (Fig 3A, 3B.). Percutaneous coronary intervention (PCI) was performed with drug eluting stent at each coronary artery. He was defined as definite FH by the Dutch criteria (9 points) and probable FH by the Simon Broome criteria (fulfilled criteria a and e). We administered rosuvastatin 20mg with ezetimibe 10mg and atorvastatin 80mg with ezetimibe 10mg, but LDL cholesterol were confirmed to be 107mg/dL and 134mg/dL, respectively, and did not reach the target goal of less than 55mg/dL. We added PCSK9 monoclonal antibody, evolocumab (Repatha ®) 140mg/2weeks and LDL cholesterol decreased to 51mg/dL. Later, he was found to have LDLR on 19p13.2 mutation in gene study, diagnosed as heterozygous FH. FH is often underdiagnosed, has poor response to lipid-lowering drugs, and often leads to premature CVD. FH should always be kept in mind in the treatment of patients with drug-refractory hyperlipidemia.