Diffuse xanthomas with premature atherosclerosis: A case report

A 20-year-old man presented with a 2-year history of exertional chest pain with New York Heart Association functional class III–IV. At the age of 5 years old, he noticed the appearance of multiple xanthomas, which increased in size with age. His family history was unremarkable. Physical examination revealed diffuse and flat xanthomas on his neck, elbows, and buttocks and large tuberous xanthomas on his shoulders, elbows, and buttocks (Fig. 1a-1d). He also presented with bilateral upper and lower eyelid xanthelasma (Fig. 1e) and corneal arcus (Fig. 1f, white arrow). Laboratory tests revealed severe hypercholesterolemia: total cholesterol level, 714 mg/dL (18.49 mmol/L); low-density lipoprotein cholesterol level, 681 mg/dL (17.61 mmol/L); high-density lipoprotein cholesterol level, 21 mg/dL (0.54 mmol/L); and normal triglyceride level, 61 mg/dL (1.58 mmol/L). Coronary angiography revealed severe multi-vessel disease, including: critical ostial stenosis of left main coronary artery (Fig. 2a); severe stenosis of left circumflex artery with distal collateral circulation to the right coronary artery (RCA) (Fig. 2b, black arrow); and ostial sub-occlusion of RCA with whole length of diffuse and severe stenosis (Fig. 2c).

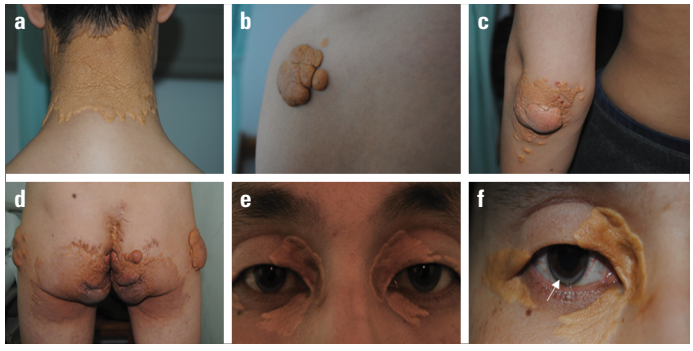

Figure 1. Diffuse xanthomas and corneal arcus

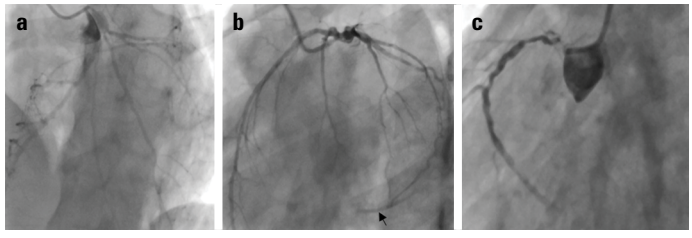

Figure 2. Diffuse, multivessel stenosis

The diagnosis of a familial hypercholesterolaemia is made on the basis of familial history, biochemical findings and genetic testing. However, this patient had an unremarkable family history of hypercholesterolemia or atherosclerosis. Accordingly polygenic hypercholesterolaemia is the most likely diagnosis, which means it might be due to a combination of multiple genetic and environmental factors. Furthermore we hypothesized that xanthomas share pathophysiological pathways with atherosclerosis in both familial and polygenic hypercholesterolaemia. The presence of xanthomas can indicate a more severe cardiovascular disease risk.

Suxuan Liu, Feng Chen, Yongwen Qin, Xianxian Zhao
Department of Cardiology, Changhai Hospital, Second Military Medical University; Shanghai-*China*

Address for Correspondence: Xianxian Zhao, MD,
Department of Cardiology,
Changhai Hospital,
Second Military Medical University;
168 Changhai Road 20043
Shanghai-*China*
Phone: 861 381 705 37 03
E-mail: chcardiovasology@foxmail.com

DOI:10.14744/AnatolJCardiol.2018.46504