Extensive xanthomas with severe coronary artery disease in a young patient with familial hypercholesterolemia

Vikas Agrawal* and Bhupendra Kumar Sihag
Department of Cardiology, Institute of Medical Sciences, Banaras Hindu University, Varanasi 221005, India
* Corresponding author. Tel: +91 9453300628, Email: vikky25@yahoo.com

A 20-year-old female presented to us with recent onset unstable angina. She had a strong family history of premature coronary artery disease (two elder sisters dying at a young age due to CAD). She had large tendinous xanthomas over extensor tendons of metacarpophalangeal joints and patella (Panels A–C). She also had xanthelasmas, gaint tuberous xanthomas over extensor aspects of limbs and buttocks and Plane xanthoma involving the neck (Panels D–H). The ECG showed deep ST depression in all leads with ST elevation in avR (left main pattern, Panel I). Laboratory tests showed total cholesterol of 810 mg/dL (20.9 mmol/L), low-density lipoprotein cholesterol (LDL) of 733 mg/dL (18.9 mol/L), triglycerides (TG) of 195 mg/dL (2.2 mmol/L), and high-density lipoprotein cholesterol of 39 mg/dL (1.0 mmol/L). Her coronary angiography revealed severe left main disease involving ostium and mid shaft and also ostial RCA disease (Panels I, J, and K). Genetic study showed homozygotic mutation in LDL receptor gene. The patient is being planned for urgent PCI with intensive medical therapy.

Panel A: Large tendinous xanthomas over extensor tendons of metacarpophalangeal joints.
Panel B: Large tendinous xanthomas over extensor tendons of metatarsophalangeal joints.
Panel C: Large tendinous xanthomas over patella.
Panel D: Xanthelasmas.
Panel E: Gaint tuberous xanthomas over extensor aspects of elbow.
Panel F: Gaint tuberous xanthomas buttocks.
Panel G: Plane xanthoma involving the neck.
Panel H: Xanthoma involving extensor aspect of forearm.
Panel I: ECG showing deep ST depression in all leads with ST elevation in avR (left main pattern).
Panel J: Coronary angiography showing severe left main disease involving ostium and mid shaft.
Panel K: Coronary angiography showing severe ostial RCA disease.