IMPACT OF CONCOMITANT DISEASE ON CORONARY ARTERY DISEASE

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Long-term prognostic significance of diabetes mellitus according to renal function in myocardial infarction patients. The FAST-MI 2005 registry
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Aims: We sought to determine the prevalence of genetically confirmed familial hypercholesterolemia (FH) in patients with an acute coronary syndrome (ACS) and to compare the diagnostic performance of FH clinical criteria with FH genetic testing. Impact of FH genetic diagnosis at family level was also evaluated.

Methods: Genetic study of seven genes associated with FH/increased LDL cholesterol (LDLR, APOB, PCSK9, APOE, STAP1, LDLRAP1, LIPA) was performed in 103 ACS patients, aged <65 years and with a LDL cholesterol >160 mg/dl (Figure). Dutch Lipid Clinic (DLC) and Simon Broome (SB) FH clinical criteria were applied. Familial genetic screening was performed in patients with pathognomonic or possible pathognomonic mutations.

Results: Prevalence of genetically confirmed FH was 9.7% (95% CI: 5.36–16.96%; n=10). Prevalence of probable/definite FH according to DLC criteria was 26.2% (95% CI: 18.26–35.96%; n=27). The SB criteria identified 27.2% (95% CI: 19.1–36.99%; n=28) patients with possible/definite FH. DLC and SB algorithms did not diagnose 4 (40%) and 3 (30%) of the genetically confirmed FH patients, respectively. 77.8% (95% CI: 57.27–90.63%; n=21) of patients diagnosed by the DLC algorithm and 75% (95% CI: 54.78–88.57%; n=21) by the SB criteria did not show any pathognomonic FH mutation.

Cascade genetic testing in first-degree relatives prompted the identification of 10 additional FH individuals. Seven of the family carriers had already high cholesterol, although only one of them was diagnosed of hypercholesterolemia and followed lipid-lowering treatment.

Conclusion: Prevalence of genetically confirmed FH in ACS patients aged <65 years and with a LDL cholesterol >160 mg/dl is high (around 10%). FH clinical algorithms do not accurately identify FH patients. Genetic testing should be advocated in young ACS patients with high LDL to allow prompt identification of FH patients and targeted treatment.

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