PREVALENCE OF FAMILIAL HYPERCHOLESTEROLAEMIA (FH) IN ITALIAN PATIENTS WITH CORONARY ARTERY DISEASE: THE POSTER STUDY

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Background and aims: Familial hypercholesterolaemia (FH) is well-established risk factor for cardiovascular events. High levels of low density lipoprotein cholesterol (LDL-C) contribute to severe cardiac and cerebrovascular diseases. Worldwide one subject out of 250- 500 has a genetic mutation determining FH. The POSTER Study wants to evaluate the prevalence of FH in the Italian population among subjects with recent diagnosis of coronary artery disease (CAD).

Methods: Data were collected in patients with documented CAD admitted at 82Italian cardiology centers. Dutch Lipid Clinic Network (DCLN) criteria was used to defined patients with a probable or definite FH diagnosis (DCLN score ^{3} 6) and these patients underwent to a whole blood sample withdrawal to perform a genetic test based on full gene sequencing of genes involved in FH (LDLR, APOB, PCSK9, STAP1and LDLRAP1).

Results: Overall 5415patients were enrolled and the main index event was an acute coronary syndrome (ACS), either with ST-elevetion myocardial infarction (STEMI) or without ST-elevation (NSTE-ACS) (34.8% and 37.2%, respectively). Mean age was 66±11years with prevalence of men (78%) and about 40% were already treated with statins, condition that increased after the acute event (from 39.2% to 96.5%). Atorvastatin was the most used (88%), followed by rosuvastatin and simvastatin. Based on the Dutch scores the prevalence of potential FH (score ^{3}6) was 5.1%, particularly the 0.9% of them had a diagnosis of definite FH (score >8). These patients were significantly younger than patients with a DUTCH score <6(56±10 vs 66±11, t test $p<0.001$), and the distribution of LDL-C levels shows that most of them (~87%) had a LDL-C level ≥191mg/dL. The genetic test has been performed in 259patients: 37patients (14,29%) was positive for the diagnosis of FH, in 63(24.3%) patients there was a mutation in these genes, but the genetic diagnosis was defined as not-conclusive for FH. Finally, in 159(61.3%) subjects no genetic variants of the tested genes was identified.

Discussion and conclusions: Among patients with a recent CAD the prevalence of potential FH is high, so we have selected a subgroup of patients that underwent a genetic test to evaluate the presence of a causative mutation and to improve their high cardiovascular risk. Therefore an accurate identification of FH patients will help to manage correctly their high cardiovascular risk and reduce recurrent events, especially in younger patients.