Familial hypercholesterolemia in acute coronary syndrome patients: underdiagnosis in female and in young patients

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Introduction: Familial hypercholesterolemia (FH) is often underdiagnosed, particularly in female patients (P), even during hospital admission for acute coronary syndromes (ACS). The aim of this study was to apply the Dutch Lipid Clinic Network (DLCN) Criteria in P admitted for ACS and evaluate gender and age differences.

Methods: Prospective evaluation of P with ACS admitted to a tertiary center from 2005 to 2019. Data including family history and laboratory tests was analysed for the application of the DLCN criteria and results were stratified according to ACS subtype, gender and age groups (20–39, 40–59, 60–79 and ≥80 years [y]). P were followed up for 30 days for hospitalization, recurring ACS and mortality.

Results: 3811 P were evaluated, mean age 63±13 years, 28% female and mean LDL cholesterol of 125±43 mg/dL. The admission diagnosis was unstable angina (UA) in 5%, non-ST-segment elevation myocardial infarction (NSTEMI) in 27% and ST-segment elevation MI (STEMI) in 68%.

Applying the DLCN criteria, 3089 P (81%) had a score of <3 (unlikely FH), 675P (17.7%) a score of 3 to 5 (possible FH), 41P (1.1%) a score of 6 to 8 (probable FH) and 1P (0.03%) a score of >8 (definite FH). Stratifying according to ACS type: among UA, 31P (16%) had possible FH and 4P (2.1%) had probable FH. Among NSTEMI, 145P (14.2%) had possible FH, 9P (0.9%) probable FH and 1P (0.03%) definite FH. Finally, among STEMI P, 497P (19.1%) had possible FH and 28P (1.1%) probable FH. Regarding female P, 158P (14.7%) had possible FH and 16 P (1.5%) probable FH.

Among male P, 517P (18.9%) had possible FH and 25P (0.9%) probable FH (p=0.016 for interaction).

According to age groups, among P aged 20–39 y (136P), 61P (44.9%) had possible FH and 6P (4.4%) had probable FH. Concerning P aged 40–59 y (1766P), 575P (32.6%) had possible FH, 31 P (1.8%) probable FH and 1P (0.1%) definite FH. With regard to P aged 60–80 y (2122P), 80P (3.8%) had possible FH and 4P (0.2%) probable FH. Among P aged ≥80 y (1837P), only 9P (0.5%) had possible FH and no P had probable FH.

In a 30-day follow-up, there was an hospitalization rate of 3.5% (134P) and recurring ACS in 1.7% (65P), while the all-cause mortality was 2% (78P) and cardiovascular (CV) death was 1.3% (49P). Female P had a significantly lower hospitalization rate (1.8% vs 3.2%, p=0.003) as well as fewer recurring ACS (0.6% vs 1.7%, p=0.001). There was no significant gender difference regarding all-cause mortality (female 1.7% vs 1.5%, p=0.552) or CV death (0.8% vs 1.1%, p=0.323). The DLCN criteria score was significantly correlated with admission for recurring ACS (OR 1.19 [95% CI 1.04–1.36], p=0.04).

Conclusion: Application of the DLCN criteria in female P admitted for ACS revealed 158P (14.7%) with possible FH and 16P (1.5%) with probable FH. Regarding younger ACS P (20–39y), 44.9% had criteria for possible FH and 4.4% for probable FH, prompting us to do not overlook these P subgroups in daily practice and routinely assess the likelihood of FH.