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Title

Familial Hypercholesterolaemia (FH) or not? FH Coding in UK Primary Care Electronic Health Records

Priority 1 (Research Category)

Cardiovascular disease

Presenters

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Abstract

Context: Familial hypercholesterolaemia (FH) is a common autosomal genetic disorder that causes premature heart disease and is presently underdiagnosed. Early identification of individuals with FH in primary care has been recommended by national guidelines to help reduce avoidable premature heart attacks and deaths. Patients with either clinically diagnosed or genetically confirmed FH have specific FH diagnostic codes recorded in their primary care electronic health records (EHR).

Objective: To assess the validity of FH diagnosis recording in routinely collected primary care records in the UK.

Study design: Observational study

Setting or dataset: Primary care practices

Population studied: Individuals registered with a participating General Practice (14 practices within Lipid Specialist Services in Leicester and Derby), aged 18 years and older with a cholesterol record and coding for FH in their EHR.

Intervention/instrument: A genetic test for FH mutation.

Outcome measure: A genetic mutation for FH.

Results: Of 32 eligible individuals invited, 16 responded and were genetically tested for FH mutation. Of the 16 tested, the median age was 72 years (60.5-74) and 9 (56.3%) were women. The median for the highest ever recorded cholesterols (in mmol/l) were: 6.4 (5.4-8.1) for total cholesterol; 3.6 (3.0-5.4) for LDL-C; 1.8 (1.1-3.2) for triglycerides, and 1.5 (1.4-2.2) for HDL-C. At the time of highest cholesterol record, 10 (62.5%) patients were on statins, with 4 (25%) on high-intensity statins. Only 1 (6.3%) patient was found to have monogenic FH with an LDLR gene. None had a variant of uncertain significance; however, intermediate, and high polygenic scores were observed in 10 (62.5%) and 4 (25%) individuals, respectively.

Conclusions: Although many individuals with FH remain undiagnosed, a higher proportion of patients with coding for FH in the UK primary care setting do not have a genetic mutation for FH. With the rollout of FH genetic testing, patients with an FH coding in primary care, based on clinical phenotype, should be offered genetic testing to confirm the diagnosis. Based on our study findings, a higher proportion of patients with FH diagnostic code in EHR will have polygenic hypercholesterolaemia.