Familial Hypercholesterolemia (FH) Community Initiative Using Electronic Health Records (EHR)

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Background/Synopsis: Familial hypercholesterolemia (FH) remains a significantly undertreated disease in the U.S. despite the availability of effective therapies. Without intervention, patients face high morbidity and mortality rates; however, early diagnosis and treatment to guidelines can reduce event rates to those found in the general population.

Objective/Purpose: We sought to implement an integrated initiative to increase early identification of and therapy for FH patients in Lancaster County. Our objective was to electronically identify potential FH patients using NLA guidelines and contact them via letter while simultaneously raising awareness of the condition among affiliated medical providers.

Methods: We queried the community hospital Epic® EHR for patients having at least one encounter at Lancaster General Hospital (n=792,752) that met the following parameters:
1. Visit with a primary care provider since 2013
2. LDL cholesterol ≥190
3. FH indicated on problem or diagnosis list.

Letters were sent to patients identified with these criteria. The letters notified patients of their elevated cholesterol levels and the possibility of a genetic component to this elevation. Each patient was tagged in the EHR to increase visibility to all providers. Providers received system-wide communiqués about the program. We evaluated our patient cohort before and six months after receiving the letter by examining the number of patients with FH added to their problem or diagnosis lists and/or started on a lipid-lowering medication.

Results: There were 3,388 patients identified with the above parameters and a cohort numbering 2,060 were contacted. Before the letter 12.6% of the cohort had FH on their problem list, 13.7% had FH on their diagnosis list, and 84.4% of these patients were on an FH medication. 11.7% of the cohort met all three criteria. After receiving the letter, there was an increase to 16.7%, 17.2%, and 86.6%, respectively. The percentage meeting all three criteria increased to 15.4%.

Conclusions: Our novel direct outreach method was successful in increasing the identification of FH in our community. We will use insights from this study to further investigate provider and patient outreach. Optimizing these procedures could provide a model approach to FH care for other community hospitals.


Diagnosing Familial Hypercholesterolemia (FH) in the United States: Results from the CASCADE FH Patient Registry

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Background/Synopsis: Familial hypercholesterolemia (FH) is an autosomal dominant disorder with a prevalence of 1:300 to 1:500 worldwide. FH patients have 20-fold increased risk for premature coronary heart disease (CHD) due to lifelong extreme elevations in LDL-C. Three criteria can be used to diagnose FH: Simon-Broome, Dutch Lipid Clinic Network (DLCN) or United States (US) Make Early Diagnosis to Prevent Early Deaths (MEDPED). In the Netherlands the DLCN criteria helped to identify 71% of estimated cases for genetic testing, early treatment, and CHD prevention. In the US, however, < 10% of FH patients are identified, perhaps due to a lack of a nationwide consensus on diagnostic criteria.

Objective/Purpose: To characterize the FH diagnostic criteria applied by US lipid specialists participating in the FH Foundation’s CASCADE FH (CAscade Screening for Awareness and DEtection of Familial
Hypercholesterolemia) patient registry — a U.S. registry that became active in September 2013 and currently has data on FH patients treated at 11 specialty lipid clinics.

**Methods:** We queried the CASCADE FH database for the diagnostic criteria chosen for each patient and baseline patient characteristics. Diagnostic criteria were divided into five non-exclusive categories: “clinical diagnosis,” MEDPED, Simon-Broome, DLCN, and/or other. Only adults (age >18) were included since the diagnostic criteria cannot be applied to pediatric populations.

**Results:** There were 876 FH adults who had available data entered from clinical sites. Fifty-seven percent were female; mean (SD) age 53 (17) yrs; and BMI 28 (6) kg/m2. Ethnicity/race was 78% white, 6% African-American, 3% Hispanic, and 12% other. Mean age at FH diagnosis was 43 (19) yrs. Thirty-eight percent had prior CHD, 16% had tendon xanthomas, and 45% had a family history of myocardial infarction. Mean pre-treatment LDL-C was 269 (87) mg/dL. Most adults enrolled in CASCADE FH received a “clinical diagnosis” of FH: 64% “clinical diagnosis” (n = 560) vs. 11% MEDPED (n = 105) vs. 4% Simon-Broome (n = 32) vs. 1% DLCN (n = 7) vs. 1% other (n = 9) vs. 19% “multiple diagnostic criteria” (n = 163), p = 0.01.

**Conclusions:** Among U.S. lipid clinics participating in the CASCADE FH registry, most did not report utilizing one of the existing diagnostic tools. Our findings imply that established FH criteria are not regularly utilized to diagnose FH in the U.S. A need exists to develop a nationwide consensus, which will lead to better identification, earlier treatment, and ultimately prevention of CHD events.

**Objective/Purpose:** To characterize the demographics, clinical parameters, and treatments of patients with LDL-C \(\geq 190\) mg/dL.

**Methods:** We performed a single center, retrospective descriptive study. Patients were identified if they had an LDL-C \(\geq 190\) mg/dL and/or non-HDL-C \(\geq 220\) mg/dL on a lipid panel performed between July 1, 2011 and June 30, 2012, at Thomas Jefferson University Hospital. Data were recorded for the 12-month period following the qualifying laboratory test.

**Results:** Of the 29,095 patients with LDL-C levels, 735 (2.5%) met lipid inclusion criteria, with a mean qualifying LDL-C of 214.2 mg/dL. The mean age of the population was 54.8 years, 61.8% were female, and 44.9% self-identified as White. Hypertension was diagnosed in 53.2% of patients, 21.8% were diagnosed with diabetes, and 18.9% were current smokers. Cardiovascular disease (CVD) was present in 19.1% of patients, and 49.4% had a family history of CVD. At the time of the qualifying LDL-C level, lipid lowering therapy (LLT) was already recommended in 33.6%. During the study period, of the 637 patients with follow-up data in our electronic medical record system, statins alone or in combination with other LLT was recommended for 58.2% of patients, and 21.5% of these patients received a high-intensity statin. Of those newly initiated on LLT with available follow-up LDL-C values, 13% of patients achieved \(\geq 50\%\) LDL-C reduction, and 24.3% achieved a 30-49% reduction. Family medicine physicians cared for 46.1% of the population.

**Conclusions:** Patients with LDL-C \(\geq 190\) mg/dL comprise a small, but significant, proportion of patients within our single hospital system. Although these data were collected prior to the 2013 ACC/AHA guidelines release, only 58.2% were prescribed statins, and of those, less than one in four was prescribed guideline recommended high-intensity statin. These patients also had significant comorbidities, and were largely cared for by primary care providers. Education of providers on the implementation of the 2013 ACC/AHA cholesterol treatment guideline could improve care of this high-risk patient population.

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**Background/Synopsis:** The 2013 ACC/AHA blood cholesterol guidelines identified four groups for whom cholesterol treatment with statin therapy is recommended to reduce atherosclerotic cardiovascular disease (ASCVD) risk. Two of these groups, those with clinical ASCVD or diabetes, have well documented clinical characteristics and outcomes trials using statins; however, the group with low density lipoprotein cholesterol (LDL-C) \(\geq 190\) mg/dL, has not been studied as rigorously. Understanding who comprises this group, their comorbidities, and their healthcare utilization patterns are of importance given the focus on this population by the recent ACC/AHA guideline.

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