Implementation of Flag, Identify, Network, Deliver: FIND FH® Using the Electronic Medical Record to Identify Familial Hypercholesterolemia within a Single Healthcare System

Samip Sheth*, Latoya Williamson, Jinbo Chen, David Staszak, Gayley Webb, Marjorie Risman, Yuliya Borovskiy, Erik Hossain, Jihane Hajj, Kady Diu, Julia Larsen, Jennifer Boring, Cynthia Mays, William Howard, , David Zuzick, Daniel Soffer, Marina Cuchel, Samuel Gidding, Katherine Wilemon, Daniel J. Rader, Kelly Myers, Douglas Jacoby

University of Pennsylvania, Philadelphia, PA
FH Foundation, Pasadena, CA
Quest Diagnostics

Background

Familial hypercholesterolemia is a common underdiagnosed and undertreated condition that leads to premature cardiovascular disease. The FH Foundation developed a machine learning algorithm (MLA) to identify at-risk individuals for targeted screening for FH. FIND FH was implemented to evaluate the likelihood of patients with a Probable FH score having an FH clinical diagnosis or FH-causing mutation in the preventive cardiology clinic of the University of Pennsylvania Healthcare System (UPHS).

Methods

A list of patients was identified by the FIND FH algorithm in the UPHS electronic medical record using a FIND FH score above 0.20, suggestive of 'probable FH,' and was then organized into three sets. For 'set 1' of FIND FH patients, the patient's primary provider was contacted to seek permission in order to contact the patient. Willing patients underwent a clinical assessment for FH. For 'set 2' of FIND FH patients, text-mining was used to determine how many patients previously seen in the clinic had "familial hypercholesterolemia" in their medical record. Genetic testing for FH was performed by Quest Diagnostics for 'set 3' of FIND FH patients, a subset of patients from sets 1 and 2.

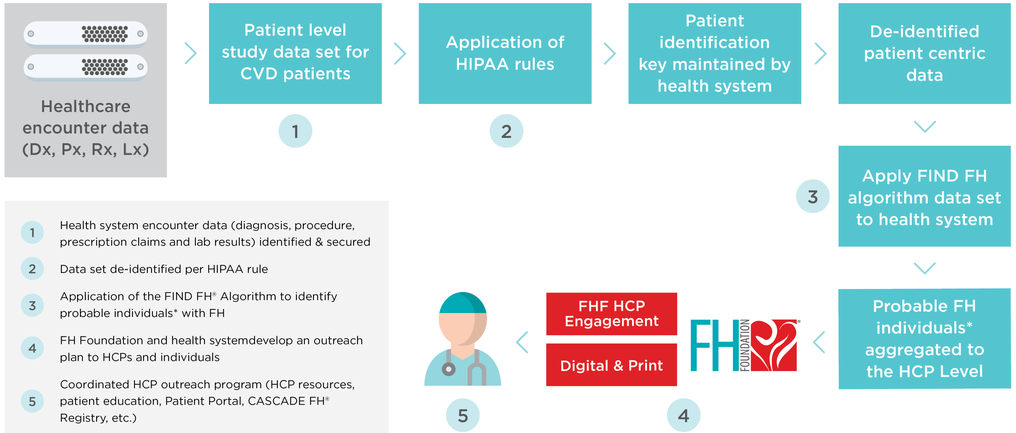
Results

A total of 8614/1.6 million individuals were identified by FIND FH. In set 1, 67 of these were clinically evaluated. Median age (IQR) was 55 (44-65), 28 (42%) were female, and 49 (73%) were white. Of these, 26 (38.2%) had possible, probable, or definite FH by clinical diagnosis. In set 2, 874/8164 were existing preventive cardiology patients. Of these, 310 of 874 (36%), had an FH diagnosis in their medical record. In set 3, the number of FIND FH patients with genetic testing for FH was 103; 22 (21%) had a pathogenic FH mutation while 5 (4.8%) and 2 (1.9%) others had a variant of unknown significance and a hypocholesterolemic variant, respectively.

Conclusions

In this clinical application of FIND FH, a high percentage of patients seen in the preventive cardiology clinic were recognized to have a clinical diagnosis of FH and/or a positive FH-causing mutation. Further research to assess the utility of MLA to improve the diagnosis rate, treatment patterns, and cardiovascular outcomes of FH is needed.

Figure 1: Implementation of FIND FH® in the University of Pennsylvania Healthcare System



*Patient that has a profile consistent with other individuals with FH

Contact
The FH Foundation
959 E. Walnut street Suite 220
Pasadena, CA 91106
(626) 583-4674
FINDFH@thefhfoundation.org