Initial Results from the CASCADE-FH Registry: CAscade SCreening for Awareness and Detection of Familial Hypercholesterolemia

Emily C. O'Brien[1], Emil M. deGoma[2], Patrick M. Moriarty[3], MacRae F. Linton[4], Michael D. Shapiro[5], P. Barton Duell[5], Christie M. Ballantyne[6], William A. Neal[7], Zahid S. Ahmad[8], Danielle Duffy[9], Lisa C. Hudgins[10], Linda C. Hemphill[11], James A. Underberg[12], Karol E. Watson[13], Samuel S. Gidding[14], Seth J. Baum[15], Katherine Wilemon[7], Dave Pickhardt[7], Iris Kindt[7], Daniel J. Rader[2], Matthew T. Roe[1], Joshua W. Knowles[16]

[1]Duke Clinical Research Institute, Durham, NC; [2]University of Pennsylvania, Philadelphia, PA; [3]University of Kansas, Kansas City, KS; [4]Vanderbilt University, Nashville, TN; [5]Oregon Health and Science University, Portland, OR; [6]Baylor College of Medicine, Houston, TX; [7]FH Foundation, South Pasadena, CA; [8]UT Southwestern, Dallas, TX; [9]Thomas Jefferson University, Philadelphia, PA; [10]Rogosin Institute, New York, NY; [11]MGH Heart Center, Boston, MA; [12]NYU Langone Medical Center, New York, NY; [13]UCLA School of Medicine, Los Angeles, California; [14]Nemours DuPont Hospital, Wilmington, DE; [15]Preventive Cardiology, Boca Raton, FL; [16]Stanford School of Medicine, Stanford, CA

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Background

- Familial Hypercholesterolemia (FH) is a common genetic disease that leads to substantially elevated levels of low-density lipoprotein cholesterol (LDL-C)
- The 2013 ACC/AHA guidelines for treatment of Blood Cholesterol to Reduce ASCVD risk in adults denote an LDL-C level > 190 mg/dL (suggestive of FH) as an independent high-risk feature.
- Individuals with FH have a 20-fold increase in risk of early cardiovascular disease.
- Despite its prevalence, FH remains largely underrecognized: an estimated 1.5 million individuals in the U.S. have FH, yet fewer than 10% have been formally diagnosed.

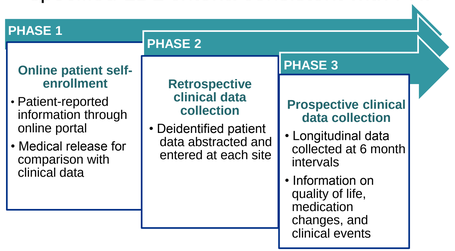
The CASCADE FH Registry

- In 2013, the FH Foundation (a patient-led nonprofit organization) created the CASCADE-FH Registry.
- The CASCADE-FH Registry is a national initiative to increase FH awareness, characterize trends in treatment, and monitor clinical and patient-reported outcomes over time.
- CASCADE-FH represents a collaboration between The FH Foundation, cardiologists, primary care providers, lipid specialists, and patients with FH.

Methods

- CASCADE-FH uses a multi-pathway enrollment strategy to ensure maximum availability to interested FH patients
- Registry participants may enroll in CASCADE-FH by one of two methods:

1) Enrollment by a provider in a specialized lipid clinic
2) Self-enrollment via an interactive online portal for participants meeting pre-specified LDL criteria consistent with FH.



Data Elements

- Demographics
- Medical history and laboratory values
- Current lipid-lowering therapies
- Clinical events (hospitalizations, mortality)
- Patient-reported outcomes (QOL, treatment satisfaction)

Data Analysis

- Baseline characteristics presented as percentages for categorical variables and medians with interquartile ranges (IQRs) for continuous variables

Results

Table 1. Baseline Characteristics of the CASCADE-FH Patient Population (September 2013 – February 2015).

Variable	Overall (N=1605)	Online Portal (N=215; 13.4%)	Clinical Sites (N=1390; 86.6%)
Median age, years	53.0	52.0	53.0
(IQR)	(37.0, 64.0)	(41.0, 60.0)	(36.0, 65.0)
<18 years of age	8.2	---	10.2
White race, %	81.4	92.7	79.6
Female sex, %	58.7	72.7	56.6
Formal FH diagnosis, %	97.4	80.6	100.0
Family member with FH, %	31.5	46.7	29.2
Prior MI, %	12.4	17.2	11.6
Prior PCI, %	17.0	19.4	16.6
Prior CABG, %	13.8	14.9	13.6
Diabetes	11.5	9.3	11.9
Hypertension	39.6	32.1	40.8
Current Smoker	6.2	3.7	6.6
Historical Highest LDL-C (mg/dL)	249.0 (211.0, 310.0)	290.0 (231.0, 355.0)	245.0 (210.0, 303.0)
Current LDL level (mg/dL)	143.0 (107.0, 196.0)	150.0 (118.0, 200.0)	143.0 (107.0, 196.0)

Abbreviations: MI=myocardial infarction; PCI=percutaneous coronary intervention; CABG=coronary artery bypass graft
*High-intensity statins defined as atorvastatin≥40 mg or rosuvastatin≥20 mg

Table 2. Treatment patterns of the CASCADE-FH Patient Population (September 2013 – February 2015).

Variable	Overall (N=1605)	Online Portal (N=215; 13.4%)	Clinical Sites (N=1390; 86.6%)
Currently treated with statins, %	74.1	73.0	76.0
Currently treated with high- intensity statins*, %	55.8	63.5	54.6
Number of LLT			
0	10.8	17.2	9.8
1	33.6	35.4	33.4
2	25.6	8.8	28.1
3+	30.0	38.6	28.7

*Among patients treated with statins
Abbreviations: LLT=Lipid lowering therapy

Results (continued)

Figure 1. Percent of patients reporting that they "completely understand" the following about FH*

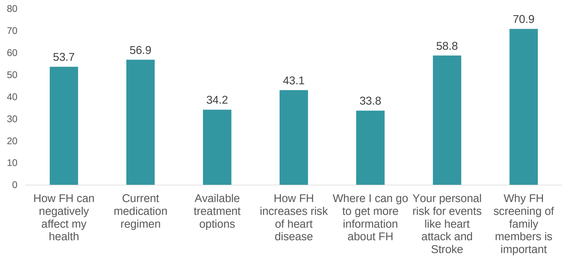


Figure 2. How satisfied are you that everything possible is being done to treat your FH?*

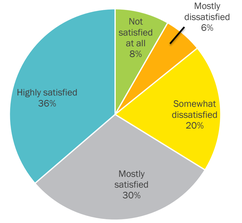
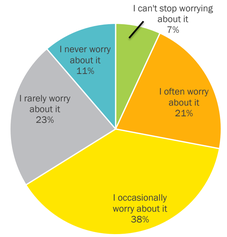


Figure 3. How often do you worry that you may have a heart attack or die suddenly?*



*Online portal patients only

Conclusions

- The CASCADE-FH Registry represents a collaboration between FH patients and clinical researchers who have partnered to address gaps in knowledge regarding FH screening, identification, and treatment.
- CASCADE-FH will evaluate future longitudinal treatment patterns and outcomes in this high risk patient population

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Contact

Emily O'Brien, PhD
Medical Instructor
Duke Clinical Research Institute
Duke Medical Center
2400 Pratt Street
Durham, NC 27705

Email: emily.obrien@duke.edu

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