DIAGNOSIS OF FAMILIAL HYPERCHOLESTEROLEMIA: A WORK IN PROGRESS

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Background and Aim

An ICD-10 code for Familial Hypercholesterolemia (FH), E78.01, became effective October 2016 following a proposal in 2013 to the ICD-10 Coordination and Maintenance Committee by the Family Heart Foundation. The code differentiated FH from other forms of elevated cholesterol, signaling the need for differential diagnosis of a condition in which management in the first two decades of life can substantially reduce the burden of aggressive atherosclerosis.[1]

This study aims to characterize the % of FH patients diagnosed with E78.01 in an expansive, real-world US dataset.

Methods

The Family Heart Database includes diagnostic data from claims for individuals from the US screened or treated for any form of cardiovascular risk. This analysis includes 197 million people, including 22 million children, from October 2016 through June 2020.

The number of total (diagnosed + undiagnosed) FH patients within the dataset was estimated assuming an occurrence of 1:250 individuals.

Patients with FH (E78.01) were counted if the diagnostic code was applied for a single in-patient claim or at least twice, >7 days apart, for an out-patient claim.

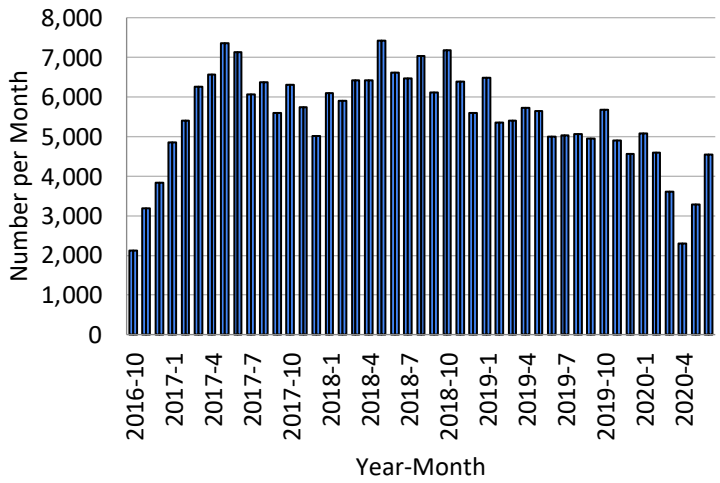

Figure 1: Number of individuals in Family Heart Database diagnosed with FH (E78.01) per month.

Results

The number of patients diagnosed with FH using E78.01 has increased substantially since 2016. During 2017 and 2018, use of the code was brisk and likely included previously and newly diagnosed individuals (**Figure 1**).

Diagnosis was reduced dramatically with the onset of the COVID-19 pandemic corresponding with the marked reduction of in-person clinic visits and near halting of preventive care (**Figure 1**).

	Family Heart Database (>197 million)	Diagnosed with FH (n=246,689)
<18 / 18-65 / >65 yr	11%/ 58% / 27%	2%/ 51% / 47%
Female	53%	52%
Black	6%	8%
Hispanic	5%	5%
White	35%	54%
Other/Unknown	53%	33%
ASCVD	8%	40%
Household Income: <$30K	9%	13%
>$100K	13%	20%

Table 1: Characteristics of all individuals in the database versus those diagnosed with FH

Results (cont.)

By June 2020, 246,689 patients were diagnosed with FH representing 31.3% of the estimated total (diagnosed + undiagnosed) FH population of 787,886 within the dataset (**Figure 2**).

Compared with all individuals in the database, those diagnosed with FH were substantially more likely to have ASCVD (40% versus 8%; **Table 1)**. Patients diagnosed with FH were also more likely to be White, and the % of Other/Unknown is high and variable in both groups.

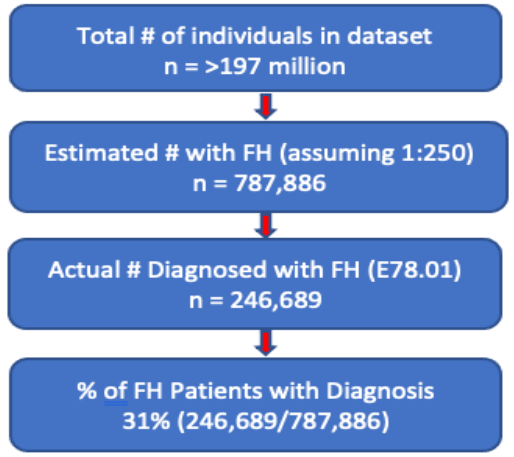

Figure 2: Family Heart Database of US individuals from Oct 2016 to Jun 2020

Discussion and Conclusion

Prior to 2016, an estimated <1% of patients with FH in the US were diagnosed [2], but without an ICD code it was impossible to track.

The number of patients diagnosed with FH (E78.01) has increased substantially since 2016. Within this large, real-world dataset of Americans, 31.3% of the estimated FH population had been diagnosed as of June 2020.

However, despite clear screening guidelines, effective therapies, and classification of FH as a public health threat by the World Health Organization[3], most of the population remains undiagnosed, leaving these genetically vulnerable individuals at high risk for premature cardiovascular disease.

[1] Luirink IK, Wiegman A, Kusters DM, et al. 20-year follow-up of statins in children with familial hypercholesterolemia. NEJM 2019; 381:1547-1556. [2] Nordestgarrd BG, Chapman MJ, Humpries SE, et al. Familial hypercholesterolemia is underdiagnosed and undertreated in the general population: guidance for clinicians to prevent coronary heart disease. European Heart Journal (2013) 34:3478-3490. [3] Representatives of the Global Familial Hypercholesterolemia Community, Reducing the Clinical and Public Health Burden of Familial Hypercholesterolemia: A Global Call to Action. JAMA Cardiol (2020) 5(2):217-229.

FAMILY HEART FOUNDATION
Lipoprotein(a) & Familial Hypercholesterolemia