Familial hypercholesterolemia (FH) is one of the most commonly occurring genetic disorders in the world. It is estimated that 1 in 250 Americans have heterozygous FH and about 1 in 160,000 to 300,000 have homozygous FH (HoFH).

Homozygotes are often unresponsive to dietary and pharmacologic intervention and often develop clinically significant ASCVD before reaching the age of 30.

Most FH patients remain undiagnosed and untreated.

The National Lipid Association (NLA) conducted a survey to evaluate the knowledge and practice of primary care and other clinicians for patients with HoFH.

These clinicians are usually the first in the healthcare community to see these patients.

Background

Methods

- An independent research and survey vendor was used to conduct the survey.
- The survey was sent out via e-mail to 14,904 medical professionals across the United States (US) from a registry of practitioners who agree to take surveys and who matched the specialty requirement.
- The survey was fielded from June 26, 2018 to July 16, 2018.
- A total of 504 clinicians completed the survey.

Screening Criteria

- US-based clinicians
- Currently treating patients with elevated LDL-cholesterol
- Licensed to prescribe medication
- Eligible medical disciplines:
  - Physician
  - Nurse Practitioner
  - Physician Assistant
- Eligible specialty groups:
  - Family Medicine
  - General Practice
  - Internal Medicine
  - Cardiology

Results - Respondent Demographics

- Over four-fifths of respondents (85%) were physicians.
  - 85% (430) Physicians
  - 8% (40) Physician Assistants
  - 7% (34) Nurse Practitioners
- Almost all respondents (99%) reported a medical area of practice in primary care and general medicine.
  - 48% (241) Family Medicine
  - 47% (236) Internal Medicine
  - 4% (23) General Practitioner
  - 1% (4) Cardiology
- The overwhelming majority of respondents do not consider themselves lipid specialists (88%).
- The majority (92%) are not certified by the American Board of Clinical Lipidology (ABCL) or Accreditation Council for Clinical Lipidology (ACCL).
- The majority of respondents (56%) practice in a suburban setting.
  - 56% (281) Suburban
  - 30% (153) Urban
  - 14% (70) Rural
- Two-thirds of respondents are in private practice.
  - 67% (338) Private Practice
  - 22% (109) Health System
  - 9% (44) Institution / Academic
  - 1% (6) VA / Government Health System
  - 1% (7) Other
Serving the Underserved: Are We Overlooking HoFH Patients?

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**Risk Assessment**
- Respondents use family history and LDL-C most often as risk factors to determine CVD risk.
- Over four-fifths (82%) of respondents used risk calculators to determine if patients were at high risk for ASCVD.

**Diagnosis and Management of Patients with an Untreated LDL-C ≥ 400 mg/dL**
53% would diagnose this as HoFH. 70% say their first step would be to **treat with medication**. Diagnosis of HoFH was not improved by knowledge of elevated cholesterol levels in all offspring.

**Very High LDL-C in Respondent Practices**
- 69% have patients with untreated LDL-C of ≥ 400 mg/dL
- 76% have patients with treated LDL-C of > 300 mg/dL
- 59% have patients who have a parent with LDL-C ≥ 400 mg/dL and all offspring with LDL-C 160-400 mg/dL

**Diagnosis with Genetic Testing and Clinical Criteria**
- The majority of respondents report diagnosing a patient with HoFH. Of those:
  - 37% (105) used genetic testing to diagnose
  - 63% (180) used clinical criteria to diagnose
Treatment Plans

- Three-quarters of respondents have a patient that was diagnosed with HoFH. Of those:
  - 64% (218) have a patient on a PCSK9 inhibitor
  - 24% (81) have a patient on LDL-Apheresis
  - 43% (217) of respondents have a treatment goal of LDL-C <100 mg/dL for those HoFH patients who are free of Clinical ASCVD.
    - 33% (168) have a treatment goal of an LDL-C decrease of 50%
    - 24% (119) have a treatment goal of LDL-C <70 mg/dL

Age at Which Respondents Would Start HoFH Patients on LDL-C Lowering Medications

The majority of respondents would start an HoFH patient on LDL-C lowering medications at 19 to 29 years of age, for both males and females (54% and 51%, respectively).

Treatment Trends

- Four-fifths of respondents would use high dose statins to treat HoFH patients.
  - 80% High Dose Statins
  - 63% Diet and Exercise
  - 60% PCSK9 Inhibitor
  - 39% Ezetimibe
  - 29% Fish oil
  - 13% LDL-Apheresis
  - 7% Lomitapide

Access to Therapies

- Two-thirds of respondents report their practice has access to a lipid specialist.
  - Almost two-thirds of respondents who do not consider themselves lipid specialists report their practice does have access to lipid specialists.
  - Less than one-third of respondents have access to an LDL-Apheresis Center.
    - Access to an LDL-Apheresis Center had a major impact on whether or not a respondent has an HoFH patient of LDL-Apheresis.
    - Access to an LDL-Apheresis Center is lacking across all regions, with approximately two-thirds of respondents not having access in each of the four US Census Regions.
    - Access to an LDL-Apheresis Center is lacking across all locations, with rural practices having the least access.

Age at Which Respondents Would Start a male Patient on LDL-C Lowering Medication?

Age at Which Respondents Would Start a female Patient on LDL-C Lowering Medication?

The age at which respondents would start a patient with HoFH on LDL-C lowering medications remains consistent regardless of the respondent’s medical specialty.
Many clinicians:
- Do not recognize HoFH.
- Do not adequately treat HoFH.
- Do not have access to a lipid specialist.
- Do not have access to a LDL-apheresis center.

There is a need for:
- More education for clinicians in recognizing and treating HoFH.
- Greater access to lipid specialists.
- Greater access to LDL-apheresis centers.

References


Financial Disclosure

Supported in part by Aegerion, Inc. and REGENXBIO Inc.

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This study was developed the NLA’s Health Quality & Research Committee.

For questions, contact us at webinars@lipid.org or visit www.lipid.org

Quick EHR Search Terms*
- Untreated LDL-C ≥ 400 mg/dL
- Treated LDL-C >300 mg/dL
- Family history of high cholesterol
- Family history of premature CV disease

*These terms can be integrated into a physician’s or practice’s electronic health record to better identify patients with HoFH.

Conclusion

There is a need for:
- More education for clinicians in recognizing and treating HoFH.
- Greater access to lipid specialists.
- Greater access to LDL-apheresis centers.

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