Electronic Health Record-based Phenotyping Algorithm for Familial Hypercholesterolemia

IMPLEMENTATION CONCEPT
Flowcharts

Authors and contacts:

Principal Investigator: Iftikhar Kullo, MD Kullo.Iftikhar@mayo.edu

Adelaide Arruda-Olson, MD, PhD ArrudaOlson.Adaleigh@mayo.edu
Carin Smith Smith.Carin@mayo.edu
Hongfang Liu, PhD Liu.Hongfang@mayo.edu
Majid Rastegar Mojarad.Majid@mayo.edu
Maya Safarova, MD, PhD Safarova.Mayya@mayo.edu
Parvathi Balachandran, MBBS Balachandran.Parvathi@mayo.edu
Saeed Mehrabi Mehrabi.Saeed@mayo.edu
Sungjhan Sohn, PhD Sohn.Sungjhan@mayo.edu
Xiao Fan, PhD Fan.Xiao@mayo.edu
Yijing Cheng Cheng.Yijing@mayo.edu

Address for correspondence:
Cardiovascular Biomarkers Research Laboratory
Department of Cardiovascular Diseases
Mayo Clinic, Stabile Building, Office 4-50
200 First Street SW, Rochester, MN 55905

Modification of a version 1.0 from May 11, 2015

Mayo Clinic Algorithm for Identification of FH from EHR

SEARCH eAlgorithm for FH version 2.0 from June 2016
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STAGE I: identification of a case-control status for primary hypercholesterolemia

Identify individuals >=18 years old with lipid profile

Extract data for all LDL-C and TG measurements

TG >=500 mg/dL on >=2 occurrences

Yes → Exclude patients

No → Identify date of the highest LDL-C level (Index date)

Extract associated age, gender, race, lipid profile

Secondary causes of hypercholesterolemia within 1 year prior to the index date

Yes → Exclude patients

No → Extract data on lipid-lowering treatment within 1 year to 6 weeks prior to the index date

LLT

Yes → Calculate a pre-treatment LDL-C level: divide index LDL-C by 0.7

No →

Cases: LDL-C >=155 mg/dL

Controls: LDL-C <=130 mg/dL

Unknown: LDL-C {131 - 154}

LLT=lipid-lowering treatment, LDL-C=low-density lipoprotein cholesterol, TG=triglycerides
Stage II: identification of a case-control status for familial hypercholesterolemia

Cases from Stage I

Group I: LDL-C, mg/dL
- >325 [8]
- 251-325 [5]
- 191-250 [3]
- 155-190 [1]

Group II: Personal History
- Premature CHD [2]
- Premature CVD/PAD [1]

Group III: Family History
- Premature ASCVD [1]
- Hypercholesterolemia [1]

Group IV: Physical examination
- Tendon xanthomas [6]
- Early-onset corneal arcus [4]

\[ \Sigma \text{ [highest score per each group]} \]

>=6 points → Cases
3 - 5 points → Unknown
1 - 2 points → Controls
Input to the eAlgorithm for familial hypercholesterolemia

Structured data
- Lipid profile → LOINC (Table 1)
- Secondary causes of hypercholesterolemia → LOINC (Table 2A), ICD codes (Table 2B)
- Lipid-lowering treatment → RxNorm codes (Table 3A), ICD/CPT codes (Table 3B)
- Family history of hypercholesterolemia → PPI (Table 5D)
- Personal history of premature ASCVD → ICD/CPT codes (Table 4)

Unstructured data
- Algorithm 1 → Family history of premature ASCVD
- Algorithm 2 → Family history of hypercholesterolemia
- Algorithm 3 → Personal history of premature CHD
- Algorithm 4 → Personal history of premature CVD/PAD
- Algorithm 5 → Physical exam findings of tendon xanthomas
- Algorithm 6 → Physical exam findings of early-onset corneal arcus