Introduction

• Family Health History (FHH) is a valuable and potentially low-cost tool for risk assessment and diagnosis in patient-centered healthcare.
• FHH-based risk assessment is not consistently used in primary care [1].
• Core data set needed to calculate FHH-based disease risk is not known.
• Data needed for calculating risk is not readily available, accurate, complete, or in a format that can be automatically processed by a computer.

Objectives

1. Review and analyze FHH-based risk assessment algorithms (RAAs) for a set of prevalent conditions that are genetically predisposed.
2. Identify core set of data elements required to compute the disease-specific risk based on the RAAs. The set of data elements may be used to guide and enhance the data collection process by EHR systems.
3. Classify the RAAs based on predefined criteria. This classification aims at providing guidance for prioritizing the implementation of RAAs.

Methods

• Searching the following sources for FHH-based RAAs:
  - English-language resources in MEDLINE
  - Disease-specific organizational websites
  - Commercial EHR vendors
  - Experts in the field
  - FHH-based RAAs for Cardiovascular Disease and Colorectal/Colon/Rectal Cancer

• Categories for Classification of Data Elements:
  - Demographic
  - Clinical
  - Behavioral
  - Familial
  - Genetic
  - Environmental

• Factors used for Classification of Algorithms:
  - Output Type
  - Level of Familial Association
  - Patient Cohort
  - Formal Validation
  - Algorithm availability

• Compare the core set of data elements to the standard dataset requirement given by the American Health Information Community (AHIC) workgroup.

Results

Analyzed 14 algorithms (9 from MEDLINE, 3 from experts, 2 from disease-specific websites, none from vendors).

Core Data Requirements

• 3 demographic elements (age, gender, ethnicity)
• 16 clinical data points: 3 unique to cancer, 7 unique to CVD, 6 were shared
• Behavioral data elements focused on habits, intake of food and supplements, OTC medications
• Familial data focused on the degree of relationship of family members affected by disease
• Only 1 algorithm used genetic testing data
• 2 algorithms used environmental factors

Classification of Algorithms

• Output Type: 7 algorithms provide a numeric risk, 3 give a scale and 4 give both types of output
• 69% of algorithms focused on first degree relatives and only 15% took into account first, second and third degree relatives
• Patient cohort information is not available for all algorithms
• Only half of algorithms are externally validated
• Algorithm availability is fair, but only one is available as a web-service

Only half data elements covered by AHIC.

Discussion

• Core data requirements required to compute risk, overlap between different algorithms
• Most of the demographic, clinical and genetic data can be extracted from patient records in EHR systems
• Familial, behavioral and environmental data will need to be collected from the patients
• Genetic testing data is not commonly used in RAAs
• AHIC recommendations account for a subset of core data elements
• RAAs we analyzed can be implemented as standards-based, shareable web services

Conclusion

• We found a core set of data elements required by most FHH-based RAAs we analyzed. Not all data are found in EHR systems and would need to be collected from patients. Only one algorithm is readily available as a shareable web service.
• Future research will focus on formal mapping of the core data set we have identified and corresponding consolidated CDA data elements, defining a family history data model and extending review and analysis to other diseases.

References


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