

Usability of a phenotype builder prototype and lessons learned for the design of phenotyping tools

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Abstract

The use of electronic health records (EHRs) for the development of phenotype algorithms offers much potential, but is hindered in part by the lack of portable, standardized phenotype definitions. One solution is the creation of novel informatics tools to facilitate algorithm development. Here we develop design guidelines from potential end users of phenotyping software using qualitative methods. Results are presented as themes to inform the design of future software systems.

Introduction

Phenotyping is the process of systematic collection and analysis of phenotypic data.¹ The implementation of electronic health records (EHRs) on the national level has a potential to tremendously scale the phenotyping process for clinical research through the creation of algorithms and tools to extract meaningful information from EHRs.² However, one challenge is the lack of validated tools to author and apply standardized research phenotype algorithms across different sites and EHR systems accurately and efficiently. Towards this goal, participatory design enables designers and developers to learn user needs and preferences early in the design process through a workshop of diverse stakeholders. The results are early design ideas that can lead to the development of more accurate user requirements for research phenotype algorithm creation.

Methods

Following the process of participatory design, we invited 15 potential end users at Northwestern University and Mayo Clinic to use a prototype tool called “Phenotype Builder” prior to the design session. All participants provided informed consent to participate. Each participant was given access to the tool, two use cases and asked to create two phenotype algorithms. The use of the tool was recorded using screen capture software. After they completed the cases to the best of their abilities, all participants participated in a focus group to discuss their needs regarding the design of a future phenotyping tool.

Results

Most participants had experience with existing tools such as the Measure Authoring Tool (for creating electronic Clinical Quality Measures) and i2b2. Some had very little experience with tools, but had interest and/or experience in algorithm development. The primary usability requirements that emerged from the group were to (1) Present the algorithm in both graphical and technical form. (2) Provide a feedback system for the outcome of the algorithm. (3) Allow for flexible definitions. Since the main stakeholders of this platform are researchers, the definitions should be able to be extremely flexible for exploratory research design. (4) Provide built-in definitions. Electronic clinical quality measures usually have precise definitions of patient cohorts, however, this is less likely to be true in research. It could be useful to integrate the precise definitions from the quality-based work so that the researchers can cite directly if they want. (5) Develop detailed documentation for the platform. (6) Include search functions so that codes in standard vocabularies (ICD, RxNORM, LOINC, etc.) are searchable, and provide guidance toward which concepts are actually used.

Discussion

Participants provided requirements for the design of phenotyping tools that were related to the algorithm building activities and general usability. Developing a more useable system for building phenotyping algorithms can contribute to more robust research and clinic practice.

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References

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