Journal of Biomedical Informatics 63 (2016) 1–10

Contents lists available at ScienceDirect

Journal of Biomedical Informatics

journal homepage: www.elsevier.com/locate/yjbin

User-centered design of multi-gene sequencing panel reports for clinicians

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ARTICLE INFO

Article history:
Received 28 November 2015
Revised 11 July 2016
Accepted 13 July 2016
Available online 14 July 2016

Keywords:
Genomic sequencing
Precision medicine
Genomic medicine
User experience research
Task walkthroughs

ABSTRACT

The objective of this study was to develop a high-fidelity prototype for delivering multi-gene sequencing panel (GS) reports to clinicians that simulates the user experience of a final application. The delivery and use of GS reports can occur within complex and high-paced healthcare environments. We employ a user-centered software design approach in a focus group setting in order to facilitate gathering rich contextual information from a diverse group of stakeholders potentially impacted by the delivery of GS reports relevant to two precision medicine programs at the University of Maryland Medical Center. Responses from focus group sessions were transcribed, coded and analyzed by two team members. Notification mechanisms and information resources preferred by participants from our first phase of focus groups were incorporated into scenarios and the design of a software prototype for delivering GS reports. The goal of our second phase of focus group, to gain input on the prototype software design, was accomplished through conducting task walkthroughs with GS reporting scenarios. Preferences for notification, content and consultation from genetics specialists appeared to depend upon familiarity with scenarios for ordering and delivering GS reports. Despite familiarity with some aspects of the scenarios we proposed, many of our participants agreed that they would likely seek consultation from a genetics specialist after viewing the test reports. In addition, participants offered design and content recommendations. Findings illustrated a need to support customized notification approaches, user-specific information, and access to genetics specialists with GS reports. These design principles can be incorporated into software applications that deliver GS reports. Our user-centered approach to conduct this assessment and the specific input we received from clinicians may also be relevant to others working on similar projects.

1. Introduction

Customizing healthcare based on each person's unique genetic makeup could enable an era of precision medicine that would improve prevention, diagnosis and treatment for many types of health conditions. Routine precision medicine is rapidly approaching due to increased use of whole genome, whole exome, and other

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http://dx.doi.org/10.1016/j.jbi.2016.07.014
1532-0464/

types of multi-gene next generation sequencing panels, hereafter referred to as genomic sequencing (GS). Results from GS will be used more often as cost goes down and evidence of clinical utility increases [1,2]. With the overwhelming amounts of data that can be generated from GS, the laborious task of manually prioritizing clinically significant results often falls to clinical and laboratory geneticists [3]. With the anticipated increase in the use of GS, manual review of genetic data by clinicians, however, is not scalable. We believe that developing computerized tools to help non-genetics experts make sense of GS results will greatly increase the likelihood of achieving the vision of successful use of these data.

The healthcare environment in which such computerized tools might be deployed can be complex and high-paced. Thus, there is a need to use a design methodology that aims to support the current way of working. The primary objective of this study was to develop a high-fidelity prototype for delivering GS reports to clinicians that simulates the user experience of a final application. Target stakeholders were clinicians involved in two University of Maryland Program for Personalized and Genomic Medicine (PPGM) initiatives that are exemplary use cases for precision medicine programs more broadly. These initiatives are the Translational Pharmacogenetics Program (TPP) and the Personalized Diabetes Medicine Program (PDMP). The TPP project aims to use a patient's *CYP2C19* genotype results to tailor antiplatelet therapy after a cardiac stent has been placed [4]. The PDMP is designed to implement, disseminate and evaluate an approach to identifying and genomically diagnosing highly penetrant genetic forms of diabetes. The goal for PDMP is to enable personalized treatment for better and potentially less invasive glucose control, prognosis, and assessment of familial risk for patients [5,6].

We employed a user-centered software design approach in a focus group setting in order to facilitate gathering rich contextual information with a diverse group of stakeholders potentially impacted by the delivery of GS reports relevant to the TPP and PDMP projects. We completed two studies: The goal for the first study (phase 1) was to understand current genetic and laboratory testing processes for documenting results, notifying clinicians of those results, and viewing the results. Findings from the first study were used to propose the design of software, which we called the Genomic Medicine Assistant (GMA), for delivering GS reports to clinicians. In particular, we proposed a design that would potentially mitigate issues with the reporting and notification processes we identified in phase 1. The goal for the second study (phase 2) was to gather feedback on a proposed design of the GMA software including preferences for interactions with, content contained in and the perceived usability of the design.

2. Background

Much of the research surrounding the delivery of individual GS results to date has been regarding the ethical, legal and social implications for communicating findings to patients [7–9]. Indeed, GS technology is not mature and appropriate in all clinical scenarios and there is the potential for cascade effects (i.e., a chain of events initiated by an unexpected result leading to unnecessary additional testing or treatments) [10–13]. We are now seeing guidance for clinicians ordering, interpreting, and communicating GS with their patients [13,14], as well as clinical laboratories supporting the ordering of GS and delivery of reports that include results that are both clinically actionable and directly relevant to the patients' indication. While there are some studies exploring the potential for GS to replace traditional tests in terms of sensitivity, specificity and completeness [15], there are few studies to date investigating technologies for delivering GS reports under this new paradigm. One group has developed a web-based platform to automatically generate a clinical report, based on pre-defined templates, from raw assay results or specified diplotypes [16]. Their framework has potential to help provide consistent and reproducible reporting while also saving time by calculating diplotypes and assembling report content. Our work adds to this emerging literature by taking a user-centered design approach to explore a range of scenarios for software generating reports from GS for use by clinicians.

Related to our goal to investigate preferences for content contained in the proposed design of software are studies of the information needs of clinicians when interpreting GS. These studies serve as the groundwork to inform the design of software tools [17,18]. While most of these studies have focused on genetic counselors' information needs, some findings may be generalized to other clinicians. Genetic counselors have expertise when reviewing GS reports; therefore, information that they deem necessary should be emphasized when non-genetic expert clinicians are reviewing GS reports. For example, a study surveying genetic counselors showed that there is a perceived lack of information on the classification system for variants of unknown significance (VUS) included in laboratory reports, highlighting a need for improved GS reports to provide transparency [18]. That study also found that genetic counselors wanted more information, such as patient information, in reports to help contextualize VUS results [18]. We believe this need for more information about the patient within laboratory reports can be generalized to other types of clinicians in order to make patient-specific recommendations. This belief is supported by findings of others indicating that clinicians desire information available to discuss with the patient [19]. Another study of interviews with genetic counselors identified specific information needs for risk communication [17]. Information needs described in that study include clinical patient characteristics, social and cognitive patient characteristics, and patient motivation and goals for the genetic counseling session. Those needs may also generalize to non-genetics experts communicating GS results to patients more broadly, and thus were considered with findings from phase 1 studies in our proposed software design.

While there exist tools and resources for clinical laboratory professionals interpreting and reporting genetic test results, the design and purpose of these tools varies. One web-based application, SeqReporter, has been developed for clinical molecular laboratory use for next-generation sequencing data analysis [20]. That tool is an automated web-based application for GS result classification. While it was designed to optimize laboratory reporting, it did not support the anticipated information needs of non-genetics expert clinical end users. While the published information provides a starting point, our work gathers additional end-user input on preferences for interactions with, content contained in and the perceived usability of the software design by non-genetics clinician experts.

2.1. Methodological background

In order to gather input on the proposed design of software, we used a task-centered system design (TCSD) protocol in a focus group setting. Other studies have used similar task-centered approaches to investigate the usability of genetic data interpretation software. For example, Shyr et al. administered surveys, conducted interviews and performed cognitive task analyses to assess the usability of a clinical exome analysis software [21]. In another study a think-aloud, graded-task protocol was used to evaluate the GeneInsight Suite. That study highlighted a need to provide the most current genetic information [22]. The graded task protocol facilitated identifying design improvements that could be easily made and determining larger issues with how the interface