Author's response to reviews

Title: Practical considerations to guide development of access controls and decision support for genetic information in electronic medical records

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Version: 2 Date: 7 September 2011

Author's response to reviews: see over
Dear editors,

We appreciate the opportunity to revise our paper, and have taken the reviewer’s suggestions into account, reformatting the paper quite significantly. We have responded to the comments as listed below in an attempt to refocus the manuscript into a more useful and meaningful paper. We look forward to your response.

Because the revisions are so substantial, text that has been changed is highlighted in two colors: green represents new text and yellow represents text that has been moved but is substantially the same. Text that has been deleted is not indicated but can be reinserted and marked if desired.

Response to major compulsory requirements

First, we thank the reviewer for his helpful and extremely thoughtful suggestions both on the structure of the manuscript and on the need to better focus on how health information technology (HIT) can be used to foster and improve use of genetic information in clinical care. We agree both that there are many unaddressed issues in terms of how to best use and incorporate genetic information into clinical care and HIT systems, and that the manuscript needs to better focus and integrate points to help the reader. To address what we interpret as the reviewers’ major concerns we have substantially restructured the paper to bring forward and add to our discussion of the characteristics of genetic information that need to be considered to guide development of HIT to facilitate adoption and patient-centered use of genetic testing results. We have tried to focus our discussion of issues around genetic information and characteristics of genetic tests around the implications for developing access controls and clinical decision support for genetic testing applications in clinical care, and attempt to make recommendations to guide HIT design where possible. As suggested, we attempt to highlight the potential benefits of electronic medical records (EMRs) for clinicians who may access and use genetic test results in clinical care situations. We have also changed the title of the paper to better reflect this re-focusing.

Response to major and minor compulsory requirements

1. Abstract. Background. The third sentence addressing variation in genetic tests, while factual, does not seem relevant and is not unique to genetic tests. The overall structure of the background in setting up the purpose of the paper was not clear.

2. Abstract. Discussion. The first sentence – this does not imply any feature of genetic tests that makes these issues more relevant than other types of lab information, or health information. Further, in the last sentence of this section regarding the role of the health care provider – this too is true for virtually all health care decisions. Unclear as to how this goes toward the relevance of the manuscript.

3. Abstract. Summary. It would be helpful somewhere to describe that this is a review of several key emerging issues pertaining to access and privacy of health information.

We deleted the suggested sentences and the discussion section and reframed the abstract in an attempt to address these points, including adding the summary requested in point 3. We refocused the abstract (page 2) and manuscript on design considerations for the development of HIT that are relevant to both access and privacy, and to guiding clinical care decisions using genetic information.

4. Background. Para 1. Sentence 5 – please clarify the meaning of “Many patients with earlier than typical cancers.....” This sentence should be referenced if stated as factual.

We clarified that we meant patients under age 50 and added a reference (see page 3).
5. Background. Para 2. Last sentence. “It is crucial to realize...” This statement may be true, but it is not unique to genetic testing results, and the relevance to the discussion in this section. This is a factor that deals with the strength of clinical evidence for supporting clinical utility and from the discussion, it's not clear how that relates to access and privacy issues of lab data in EHRs.

We have reframed this point and deleted the sentence that is mentioned above.

6. Background. Para 3. The concluding sentences, while helpful

We are not entirely sure what the author was trying to suggest since this thought was cut off, but we chose to delete several sentences in the Background section and move others to the start of the discussion section, again in an effort to better frame the paper.

7. Discussion Section A. Some context setting with regard to the connection of the types of genetic tests to privacy and access should be provided. It is difficult in most of the examples to see how the difference in types of tests has an influence on either of these two characteristics. If that is the case, then it should be easier to consolidate and summarize the types of tests and examples. If the authors are attempting to highlight distinguishing privacy and access characteristics, then a case should be made that does that. Otherwise, much of the detail here could be more easily cited to other reviews.

We have reworked this section throughout (pages 10-16), like the rest of the paper, to clarify that the type of tests have implication for who genetic information should be presented to, and the level and type of clinical decision support that will be required to make this genetic information useful in guiding clinical care.

8. Section B – Interpreting genetic tests. The discussion here seemed mostly aimed at issues involving test validity and utility. There have been many recent review papers addressing this important issue. The authors do not make it clear how these issues relate to privacy and access issues. Further, the importance of correct interpretation of test results would seem more relevant to the clinical setting of a particular patient, whereas decision support tools are more important to improve clinical care practice.

We agree with the reviewer that issues around test validity and utility are important for guiding the clinical care setting where this information should be considered and used, and the decision support tools required to effectively use this information. We have reworked this section throughout (pages 16-21) to emphasize how the issues around test validity and utility should inform when, to whom, and how health IT should present specific types of genetic testing information and when supporting information would be required.

9. Section B – The implication of simple versus complex genetic tests results and how that relates to EHRs and privacy/access considerations was not apparent. The relevance of this section, while important in understanding clinical uses of genetic tests should also address potential solutions that can be addressed through medical record systems.

We have added recommendations throughout this section (pages 16-21) for how medical record systems could handle simple versus complex genetic tests, specifically how they could be made available to different types of clinicians and with different supporting information.
10. Section B – SubSection 2. Complex tests – paragraph 5. The relevance of the first sentence is not understood. All tests have limitations. It is not clear what the point of this comment is.

This sentence (and entire paragraph) have been deleted from this section of the paper (pages 18-21).

11. Section B – Subsection 3, Para 1. The point addressing variation in lab reporting styles is not exclusive to genetic tests. One major point not addressed here is that relative to other types of lab information, there tends to be a highly developed nomenclature and standards for describing polymorphisms, and other genetic aberrancies. While genetic test literacy is a major issue, many would argue that in a comparative way, genetic test information is relatively highly structured and consistent. If the transmission of the reports and the way interpretations are provided are what is intended here, that is a different issue than reporting results and this should be clarified.

We have removed this section of the paper. Discussion of how different laboratories report results has been incorporated into Section B (page 17-18) and clarified that the transmission of reports and their interpretations by the labs is what is meant.

12. Section C. Recording genetic tests. While these points are interesting, they lack specificity that would help designers or clinicians know what would improve the situation. There is no discussion here about the structure of electronic records, transport standards, integration of problem lists and test results, etc. and these are all important points that should be considered for an informatics publication.

We have removed this section of the paper and incorporated a few minor points earlier in the paper for clarity.

13. Section D. These are relatively general statements regarding the emerging health IT infrastructure. For this section to provide new information and context, details of the components of health IT are needed, including addressing incentive structures that drive adoption and redesign of software systems to address the issues that seem to be the main feature for the paper. Further, many existing EHR systems today do not have the features desired by the authors in this session. A more useful discussion would be to provide insights into how development of future systems could be engineered to meet these needs, and the policy issues that led to the movement to higher level of functionality in HER systems.

We have removed this section from the paper and now attempt to provide suggestions throughout the paper for how future health IT systems could have specific features incorporated that would meet the needs of appropriately managing genetic test results. A few minor points from this section were incorporated earlier in the paper.

14. Section on Balance Between Privacy and Accessibility. This seems to be the major area of focus by the authors and it would be helpful to get to the arguments in this section much earlier in the paper.

We have restructured the paper to move these points earlier to the opening section of the paper and consolidated them into three primary dimensions of genetic tests that are most relevant to their incorporation into EMRs. We agree with the reviewer that it is helpful to get to these arguments earlier in the paper (pages 4-9).
Further, the discussion format here really doesn’t build on the key principles made earlier in the manuscript (see above general comments). Further, in this section, the authors chose to contrast conditions favoring one extreme (more open access vs. more restrictive). This may serve to make a point, but functionally is not really that relevant as technology designs would likely enable flexibility to accommodate many forms of access controls, personalized choice in data liquidity, etc. Throughout this section, I was looking for the authors to address interests by many patient groups to make their information more publicly available through personal health records, patient registries, and other more open access issues. The points made in this section seemed to be particularly driven by the impact on health providers and not the desires of the patient which should be included.

Throughout this new opening section (pages 4-9) we attempt to emphasize that EMRs have the potential to facilitate flexible access controls and incorporation of patient preferences into their design. We absolutely agree with the reviewer that patient preferences for either openness or restriction on their personal genetic data should be respected and hope that this is now clear in the text.

15. Section on Balance Between Privacy and Accessibility. Last paragraph. The consenting issue as described here is generalizable to all health information and is not exclusive to considerations for digital health information. The discussions on consent don’t seem to be technologically oriented or present new thinking on how technology aids (or hinders) patient authored controls for access to data.

The topic of consent forms for use of genetic test results has been substantially removed from the manuscript.

16. Summary. The set up for most of the discussion seems more related to future uses of genetic tests and not aimed specifically toward EHRs and privacy/access controls. With regard the need for future policymaking, substantial work is now ongoing with the proposal of new HIPAA regulations related to health information exchange.

The summary section has been substantially reworked to focus more on the importance of incorporating privacy and access controls and decision support capabilities in EMRs that will handle genetic test results. We feel that the topic of privacy controls in health information exchange, while important, is too broad to address in this paper.

17. Summary. Para 2. While the authors focus on the issues of mitigating risks through access controls, it does not appear that they have made a case for how genetic features require this to any degree more than other sensitive health conditions (i.e., genetic exceptionalism). The major risk would seem to be in the context of re-identifying someone using genetic test results (as a unique feature of genetic tests), but a case was not presented for this and would be highly unlikely unless large genomic sequence information was captured in an HER (very unlikely at the moment).

The restructured paper now attempts to make it clear that there are distinct features of genetic tests that are important for the designers of EMR systems to consider when structuring the system. These features do not relate to how patients view genetic information per se, but to the characteristics of genetic data and genetic test results themselves.