Reviewer's report

Title: Practical Considerations for the Accessibility and Privacy of Genetic Information in Medical Records

Version: 1 Date: 10 July 2011

Reviewer: Gregory Downing

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Biomed Central Health Services Research
Review: Practical considerations for the accessibility and privacy of genetic information in medical records. Darcy et al.
Reviewer's Report.

Major Compulsory Requirements

This paper broadly addresses issues associated with uses of genetic tests and health care applications of them with a thematic emphasis on attributes of electronic health records. This is largely a review paper and opinion piece that aims to raise awareness about pending and future issues regarding access and privacy of genetic test information. The topic matter is of growing importance to the patient care community and has grown in importance as genetic test use is increasing in practice.

Currently, the paper is organized in 2 segments: detailed discussions about genetic tests and their use, followed by discussions regarding privacy and accessibility. For the reader, this segmentation makes context difficult to relate emerging concerns on privacy and access and how they can be addressed. The integration of the policy concerns regarding privacy and access should be more closely integrated with the context of the genetic test information application. As the structure of the paper addresses the uses of genetic tests and puts emphasis on privacy and accessibility, there is little if any context given to the informatics infrastructure that supports the uses of tests, and prompts the privacy and access issues. As laid out, the authors attribute what’s different now, is the mobility of the information – thus the focus on electronic health records. The reader would likely benefit from a general sense of the issues involved with genetic tests results and decisions made with their use, but the extensive discussions in the first section are not placed in any context of how the privacy and access issues are different in the emerging world of electronic exchange of information. The early part of the manuscript should place into context a sense of how EHRs and health IT in general sets the stage for the privacy and access issues discussed later in the paper. In summary on this point, the current framework for the paper is out of balance and lacks context for the informatics framework that sets up the points for why privacy and access issues are more prominent for clinical situations involving genetic test results. Although the focus of the paper appears not to be focused on solutions, the technology capabilities
of the future will likely play a big part and the paper could be strengthened by a more detailed description of the underlying digital framework for how genetic test information is presented.

The authors provide discussions on other general points that the manuscript that are not directly related to access and privacy issues. One example of this is that there are needs for greater evidence needs to support clinical application of genetic test results. The authors did not discuss ways in which clinical information can be used to fill that gap and in turn, contribute toward improved clinical evidence. Today, this is a major theme around health IT, to support the learning health care system – wherein the clinical information supports the evidence development process that ultimately supports improved health care practices. The studies to support the clinical evidence needs for many tests are in need of more specific phenotypic information for which the electronic health records are considered to be able to support. Recently, there have been a series of published reports on the value of EHRs to support these evidence needs. It is important, relative to the context of this paper’s focus on privacy and access to point out that the maturity of both EHR systems and genetic testing application are very early in their developmental pathway toward supporting health care processes. It would have been helpful for the authors to consider the potential value that EHR systems provide as opposed to a solitary focus on potential harm as the reader may then appreciate the balance that needs to be reached from the vantage point of the IT application. Many clinicians are not yet working in EHR environments and most EHR systems are largely not able to support detailed genetic test laboratory reports in a structured format. So, in that context, the privacy and accessibility issues are not experience by a large sector of the health care enterprise. On the other hand, the issues of privacy and accessibility have been mostly a health policy focus on uses of such information by payers and insurers and potential implications that has on individuals. Here the electronic considerations for potential negative consequences to be incurred by the patient are more common and potentially implicating, even in the light of recent federal regulations for protections against certain discriminatory actions take using genetic test results. Another IT issue to consider is that while electronic transmission of health information is certainly becoming more common, the interoperability issues and ability to exchange digital health information is still very early. The implication for this point is that it is very hard to move any type of data across systems thus limiting the privacy issue to a local one, largely. At this point, there are no scientific publications that address large-scale movement of genetic test information across EHR systems.

On the policy side, the authors lightly touch on pertinent health policy uses regarding controlled access. These discussions did not shed new light or provide new avenues of research that underpin greater accessibility to information in situations where warranted and greater privacy controls. This is the subject of considerable applied research in the health IT community and probably deserves more details than being identified simply as issues, such as case-specific, role-based access; uses of authentication and auditing controls for identifying access. A more balanced approach to how the information needs for research in the paper coupled with the concerns for privacy and access would be helpful to
the reader’s comprehension about the inherent tension.

The authors, while focusing on the implications of information and potential harms from it, did not engage in any discussion about the utility of the lab results in the EHR setting. Without doubt, clinical scenarios such as many of those presented in the paper have many possible permutations of medical action. To that end, presenting options for lab ordering in CPOE functions, clinical alerts, templates for prevention screening, and clinical decision support functions are very important elements to discuss to guide uses of genetic test information. These features are important to enhancing the clinical utility of genetic testing and improving the quality of the patient experience through appropriate uses of them. The ability to take large population datasets, transfer the genetic test results into actionable medical advice, and apply that through EHR systems is arguably the most important challenge facing the uses of EHRs in genetic testing scenarios. A thorough discussion regarding accessibility should not pertain solely to the test result itself and the interpretation, but rather how those results are presented in the context of clinical situations. Readers, particularly those focused on workflow and clinical decisionmaking would look for this argument in a review paper on this topic.

Overall, the audience would likely appreciate a greater focus on the interface of the policy issues in the context of the technology. Further, a more balanced view on quality of care improvements and enablement of clinical genetic testing applications as a result of digital health information would make a much stronger paper. With so much of a paradigm shift occurring within the healthcare system in the means by which care is delivered, this represents an opportunity to overcome the issues presented.

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.

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.

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, its not clear how that relates to access and privacy issues of lab data in EHRs.

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

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.

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.

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.

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.

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 provide are what is intended here, that is a different issue than reporting results and this should be clarified.

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.

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 EHR systems.

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. 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.

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 aides (or hinders) patient authored controls for access to data.

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.

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 EHR (very unlikely at the moment).

Level of interest: An article of limited interest

Quality of written English: Needs some language corrections before being published

Statistical review: No, the manuscript does not need to be seen by a
statistician.

Declaration of competing interests:

I declare that I have no competing interests.