Author’s response to reviews

Title: Preparing the Next Generation of Genomicists: A Laboratory-Style Course in Medical Genomics

Authors:

Michael D Linderman (michael.linderman@mssm.edu)
Ali Bashir (ali.bashir@mssm.edu)
George A Diaz (george.diaz@mssm.edu)
Andrew Kasarskis (andrew.kasarskis@mssm.edu)
Saskia C Sanderson (saskia.sanderson@mssm.edu)
Randi E Zinberg (randi.zinberg@mssm.edu)
Milind Mahajan (milind.mahajan@mssm.edu)
Hardik Shah (hardik.shah@mssm.edu)
Sabrina Suckiel (sabrina.suckiel@mssm.edu)
Micol Zweig (micol.zweig@mssm.edu)
Eric E Schadt (eric.schadt@mssm.edu)

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Author’s response to reviews: see over
Dear BMC Medical Genomics Editorial and the Reviewers,

Thank you to the reviewers for your close reading of the manuscript and your many helpful suggestions that have helped us improve this manuscript. Please find our point-by-point responses below (with the text from the reviews in italics).

As background, we have received numerous inquiries from groups at other institutions considering implementing similar courses. Those inquiries motivated us to write this manuscript, as a Correspondence article, to communicate the kind of information that would be highly relevant to a course implementer, and of interest to the genomics community at large, but would not be a good fit for an Original Research report. This manuscript is intended to complement the research results reported from the companion study.

Correspondence articles are specified to be just 1000-1500 words. To stay within those limits, we pushed a lot of content, such as the “Lessons Learned”, to the supplemental. Both reviewers pointed to the lessons learned as being particularly informative and interesting. To make sure that section doesn’t get “lost” in the supplemental, we incorporated a brief listing of some of those lessons into the main text at the end of the introduction.

“A detailed description of the objectives and organization for PAPG, course syllabi and lessons learned, including the unexpectedly high uptake of sequencing by students, the challenges faced by students with limited informatics backgrounds in working at “genome scale” and the challenges of maintaining the distinction between genomics education and genome interpretation, are included in the Supplement.”

Specific responses:

Reviewer 1:

Major compulsory revisions:

1. As it stands alone, the manuscript does not add significantly to the body of knowledge or their own prior publications on this topic since they admit the inability to quantitatively demonstrate improved educational outcomes (and since the qualitative data is reported already). However, the supplemental materials contain a section “Lessons Learned” that would improve the manuscript if added to the text body to give the manuscript substance.

Please see above about our intent for this manuscript and the limitations of the Correspondence format. We too think that the “Lessons Learned” is an important part of the manuscript and so as described above have tried to incorporate a brief listing of some of those lessons into the main text so that readers have a better sense of what is in the supplemental.
2. The authors argue throughout the paper that the purpose of the course is to prepare the next generation of genomicists but neglect to define who those genomicists are (aside from a brief reference to “health professionals”). This manuscript would be greatly improved and add to the general body of knowledge with a demographic breakdown of interest in, enrollment, and completion of the course. Are the students primarily genetic counselors? Nurses? PhD scientists? MDs? The title reflects this ambiguity as well. There is plenty of published data that someone needs to be educated but not enough on whom to educate and how. This paper (in addition to the other previous works) describes one “how” but doesn’t address who will most benefit or to whom such education programs should be targeted.

We couldn’t agree more that the “whom” is a really interesting question around which there are a lot of different opinions. We tried to highlight some of that debate in the third sentence of the Introduction “reflecting the lively debate among and within academic centers about what material to teach, to whom, and how to do so most effectively.”

In the future, we expect that many different kinds of healthcare providers (not just physicians) and research scientists will encounter genomics data in routine practice, but in the near term we are focused on educating on the genetic counselors, laboratory and medical geneticists and research scientists who are already on the front-lines of genomic medicine. We made the following changes to more clearly express our expectations for the future and our current focus:. The first sentence now reads “There is an acute need for more effective genomics education for healthcare providers and research scientists”. And we replaced “health professionals” in the second paragraph of the introduction with “genetic counselors, laboratory and medical geneticists and research scientists”. With the above changes, the second paragraph of the introduction now reads:

“Despite the current uncertainty about the eventual practice model for genomics and personalized medicine, clinicians, nurses, pharmacists and other healthcare providers must begin preparing for a future where they encounter genomics data in routine practice[11]. We believe this preparation begins by training the genetic counselors, laboratory and medical geneticists and research scientists who will be responsible for translating genomic research into genomic medicine.”

The PAPG students are primarily genetic counseling students, laboratory and medical geneticists and PhD or MD/PhD students. Although the different student backgrounds were described in the last paragraph of the Introduction, the actual student breakdown was not included in the manuscript. We have added that breakdown for 2012-2014 to the Supplement and a pointer to that information in the fourth paragraph of the introduction, which now ends:

“PAPG enrolls 20-25 students per year; it is part of the genetic counseling core curriculum and offered as an elective to medical genetics residents, pathology fellows, medical and graduate students. The enrollment by student background is listed in the Supplement.”
**Discretionary Revisions:**

1. Line 33 – clarify the introductory sentence: for whom is there an acute need for more effective genomics education? (see comment above: nurses? Doctors? Scientists?)

As described above, we added additional detail to that sentence. It now reads: “There is an acute need for more effective genomics education for healthcare providers and research scientists”.

2. Line 40 – the authors refer to “the expert genomicists” but I am uncertain which populations they comprise. From this sentence I may assume the authors mean research scientists, but I suspect they may mean clinicians.

We replaced “expert genomicists” with “genetic counselors, medical geneticists and research scientists” to be more specific.

3. Lines 63-65 – the authors reference their hypothesis to improve educational outcomes, but this paper is not able to deliver on this hypothesis. Please re-word this sentence to be consistent with the papers findings.

As reviewer 1 points out we can’t yet confirm or refute this hypothesis. In this manuscript we specifically attempted to communicate that we couldn’t do so quantitatively, why that is and what we are doing differently in the future to enable to us to confirm/refute that hypothesis. It remains our motivating hypothesis that incorporating personal genome sequencing will improve educational outcomes for at least some students, and the qualitative data we have collected so far suggests that is indeed true.

**Minor Essential Revisions:**

1. Line 36 – reconsider word choice “vigorous,” a term that implies a physical component. Consider perhaps “contentious” or “provocative.”
2. Line 38 – reconsider word choice “uncertainty” in reference to genomics and PM. I don’t think that the disciplines are uncertain; perhaps the authors mean that the validity or utility of these disciplines are uncertain?
3. Line 53 – “next generation” in this context (and in the title) should not have a hyphen.
4. Line 113 – “if” is missing from the sentence.

Thank you for the close reading. We have made the above changes, replacing “vigorous” with “lively”, expanding the sentence is item 2 to read “Despite the current uncertainty about the eventual practical model for genomics and personalized medicine…”, removing the hyphens in “next generation” and adding the missing “if”.

**Reviewer 2:**
This is a timely and well written manuscript of descriptive work on a hands-on laboratory genomics course designed to train medical students, medical genetic residents, and graduate students. This approach to teaching genomics is unique, and the report provides necessary detail and instruction in the Supplemental Material such as including preparatory coursework, discussion of, and approaches to optionally preparing students for exclusion of regions for variant data they do not wish to learn about; offering genetic counseling; and maintaining privacy. The Methods are sound. Limitations are clearly stated. This comprehensive approach will allow others to develop a similar teaching model for medical students, etc, even genetic counselors in training. There is also a section on Lessons Learned which is quite informative.

Thank you for your comments. We too think the “Lessons Learned” will be very interesting to readers and as described above have tried to briefly list some of those lessons in the main text so that they are not “lost” in the supplemental.

*Might suggest the authors provide results from the 2013, 2014 cohorts as they allude to these results in the manuscript.*

A complete analysis of the quantitative and qualitative results from those cohorts is currently underway (5 surveys timepoint and one interview timepoint), but unfortunately would be well beyond the scope of this short Correspondence article.