Reviewer’s report

Title: Do patients and research subjects have a right to receive their genomic raw data? An ethical and legal analysis

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Reviewer: Aaro Tupasela

Reviewer's report:

The manuscript hand discusses a timely and important topic facing genomics research and its interface with the research subjects/patients that it engages with. Before publication, however, there are a few details that I think the authors should attend to to make the argument more precise and clear. I have listed these comments below:

- I found it refreshing and important that the authors took the time to define what they meant by 'genomic raw data.' I think this is an important point to make within the context of the paper. I would, however, like to encourage the authors to perhaps extend this a bit further by referring to 'Raw Data is an Oxymoron' book edited by Lisa Gitelman. After all, how re choose to 'read' the genome is by no means a self explanatory process, but rather requires a number of choices by researchers as to what is considered important to understanding disease and heredity. As such, the genome is just one 'thing' among many which has relevance to people health and how it plays out in life. The authors also at times appear to give the genome a higher status in terms of its relevance to human health as opposed to other factors such as life style, etc...

- I would suggest moving the discussion on p.9 relating to the problem of clinical validity and the possibility of mistakes and quality control in research to p.3 where there is a discussion of NGS as a research tool. Although the authors are correct in pointing out that the distinction between research and treatment can often be unclear, there is a big difference between clinically validated tests and research done without ISO standardisation (for example).

- The paper seems to take as its context Europe (at least in the legal discussion). This would be good to make clear at some point or otherwise it would have to provide more discussion of other relevant contexts, such as the US. There is a great deal of literature on the US context, so this choice may make the text too long. If the choice is to focus on Europe, then perhaps examples from elsewhere, such as 23andMe could be left out for the sake of clarity.

- The authors explain that the approach that they take draws on the perspective of egalitarian liberalism; although I tend to understand why, it might be useful to explicate why the notable perspectives have been dismissed.

- p.7, l. 11. What is the distinction between data and information. It may be useful to stick to using one term (data) as opposed to others which may introduce confusion.

- p.9, top. There seems to be an argument here that if people only had higher levels of knowledge regarding genes and genomes then there would not be this problem. The knowledge-deficit model has been shown to be problematic in many instances and I doubt it would help here either.
Although the discussion of costs associated with disseminating genomic data, I think it really distracts from the main discussion and could be edited a great deal to shorten the text. It would be enough to say that information dissemination incurs costs etc instead of getting deep into calculations.

There appears to be a grammatical issue with the sentences.

There seems to be an assumption in this argument that there is such a thing as a high quality testing tool. The challenge in my opinion is not about the quality of the testing tools, but the interpretations and clinical validity of the data more than anything else and not the quality of the tools. Perhaps this could be clarified to point that the problem is interpretation and validity more than anything else.

I realise that I am splitting hairs here, but I do not think that all patients should have access to their medical records. Patients with psychiatric conditions, for example, are one such case, but I suppose that these individuals would be considered as having limited capability of making decisions about themselves in any case.

What do the authors mean by 'consideration'? Such a word is rather loose and can mean many things. It either needs to be omitted or requires an explanation of what types of 'considerations' justify not sharing genomic data.

Recommendations: perhaps my biggest problem with the paper has to do with the recommendations in that they seem to solve the problems with the classical informed consent approach. Although I am in general agreement with the right to receive such data the idea that one simply provides information, sign a brief 'receipt' and you are on your way is somewhat problematic, since it comes down to what information you are providing. There is a great deal of literature out there on how people do not read, understand etc informed consent forms or procedures. As such, I cannot understand why it is introduced here as a solution, except as a legal exercise to limit liability. But why should this be the case if people are part of an egalitarian/liberal society where they are autonomous as has been argued. I think the whole paper comes undone at this point by assuming that the consent is what makes it all ok. Perhaps I come from a different theoretical perspective, but I do not think that IC is a solution or answer to this problem. Perhaps a better and more fruitful approach would be to question the relevance of genomic data in the first place. There are a number of good examples, such as from the US, where medical societies have listed conditions which should be reported back (being validated and actionable), leaving the rest of conditions to researchers to validate first. This approach also seems to have the most relevance and applicability at the national level in healthcare services. This way one does not end up with a spectrum of different approaches adopted by different institutions within one country or region, which would significantly contribute to inequality.

General comment: The paper takes its starting point that institutions are responsible for how genomic information is made available. The paper would become much stronger if it pointed out that a number of countries, such as Denmark and Finland are in the process of drafting legislation concerning genome information (Denmark), or already have laws in place (Finland), which cover the management of genomic information (see for example Tupasela and Liede, 2017 on the Finnish case).
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