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Commentary: Genetic contextualism and the value of a structured process

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Garrison et al offer a new conceptual approach to the long-standing discussion and debate over whether and how genetic information differs from non-genetic information.[1] Their intent, as I understand it, is to move beyond stale, dichotomized positions encompassed by the term “genetic exceptionalism,” toward a more nuanced discussion characterized by “genetic contextualism.” This appears to be a productive move, perhaps more for the analytic approach proposed than for any strikingly new conclusions reached.

I would note first that this article represents a wonderful example of “ELSI” research. The ethical, legal and social issues (ELSI) in human genetics has been a significant focus for the National Human Genome Research Institute (NHGRI) since its inception and this has been traditionally the largest source of grant funding for bioethics research and scholarship.[2] It does not appear that this paper was the direct result of NIH funding however the authors are prominent scholars in this field and come from centers with strong histories of ELSI funding. More specifically, the paper illustrates the cross-fertilization that comes from multi-disciplinary or interdisciplinary research. The authors bring tools familiar to rhetoricians to the biomedical domain, suggesting that this approach will bring more structure and clarity to debates in our field. Whether they are successful remains to be seen but the effort alone is stimulating and refreshing.

The conceptual approach imported from rhetoricians is “status theory” that structures a debate through attention to four questions in sequence. The approach, as described, moves from the articulation of facts, to definitions of key terms, to identification of values, and finally to implications for policy or other forms of generalization of findings. The claim is that this structured approach will assist in the identification of where discussions and debates begin to falter by facilitating a recognition of when different commentators are talking about different dimensions of an issue. In theory, this structured approach appears quite valuable.

The application of stasis theory to the domain of genomics is, of course, the focus of this paper. The authors offer the notion of genomic contextualism as a more nuanced way of discussing questions of genomic exceptionalism. This move raises an initial question of whether we should be speaking of contextualism as a single broad concept under which issues related to genetics or genomics will arise. That is, does the notion of “genomic contextualism” itself suggest that genomics is sufficiently different from other domains of

biomedicine that it needs its own form of contextualism? Is genomic contextualism a form of genomic exceptionalism? I will leave this question behind to see where the authors' analysis takes us.

In application of this structured approach to an active debate in the field of genomics, the management of secondary and incidental findings as one of their two examples, the authors first address the fact question. The relevant facts offered are that genetic test results exist. I found this basic fact to be uninteresting and not the sort of fact on which we need to spend much time in discussion. But there are a host of other facts that are not identified at this stage by the authors. An additional fact is that a genetic incidental finding in one individual is likely to reflect the genetic status of family members. A large portion of the debate over whether to disclose adult onset conditions within the ACMG 59 variants to parents of children who are sequenced is focused on the fact that a child with a BRCA1 variant is highly likely to have a parent with the same BRCA1 variant.[3] Data indicate that women with a BRCA1 mutation can reduce their risk of cancer and pursue early detection. Does this set of facts enter the discussion at this stage? If so, how does one decide what constitutes a fact? For many conditions in this context, the clinical validity and utility of genetic information is open to debate because data are limited and people interpret the existing data differently. A further elucidation of this analytic method should expand the discussion of what facts are relevant at this stage.

It appears in the authors' analysis that the key questions relevant to genomic exceptionalism arise in the second stage of the analysis, that of definitions. Here the attributes of heritability, visibility, and predictive power are identified as up for discussion. It is not entirely clear why these elements are a matter of definitions. Regardless, the discussion highlights similarities and differences between genomic and non-genomic tests – the crux of the debate over exceptionalism. The broad conclusion is that some attributes are shared across this divide and some are not. This is, I believe, consistent with the prevailing consensus on these questions. One relatively minor quibble I would have with the authors is that they highlight the 2003 article by Michael Green and myself on the question of genetic exceptionalism.[4] Building on the prior work by Tom Murray on this issue,[5] we took a relatively nuanced view of the differences between genetic and non-genetic information. The authors acknowledge this work yet maintain that the current debate is polarized or dichotomized between more starkly opposing positions. This may overstate the division in the field. The potential value of this paper is that it leads to a more nuanced view of the differences, based on the context of testing, but I am uncertain whether this is new territory or a buttressing of existing attitudes.

This proposed analytic approach also highlights values as an essential component. The authors rely on familiar bioethics principles in this initial explication. I look forward to an expanded discussion given the complexity and unresolved nature of many of the relevant values. For example, we might agree that the context is quite different between obtaining a BRCA1 test in a 40 year-old woman in a high-risk family compared to conducting BRCA1 testing in a fetus or embryo. What values are relevant to what tests are used to assess fetuses and embryos and whose values are most relevant? If pregnant couples in high-risk families are eager to use prenatal diagnosis to prevent the birth of children with a BRCA1 mutation,

is this a set of values that is determinative and what is the relevance of the values of clinicians and other social commentators like bioethicists? The advantage I see of stasis theory is that it welcomes the right focus for the discussion over values and, perhaps, permits commentators to address the same questions. The thorny and unsettled nature of many of those value questions in genetics remains.

The conclusions the authors draw on the case examples of incidental findings and data sharing are well-within mainstream thought on these issues. I take the value of the notion of “genomic contextualism” as primarily a process-based contribution – a new way to think through and discuss issues based on an established theoretical framework, albeit from a separate field. The lack of startling conclusions, at least for the two examples, is reassuring, not a bug. I look forward to seeing how this contribution to the field is used as ethical challenges continue to emerge in field of genomics.

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