Genetics inMedicine

www.nature.com/gim

Check for updates

CORRESPONDENCE

Response to Carlson

Genetics in Medicine (2021) 23:1169–1170; https://doi.org/10.1038/s41436-020-01095-5

We thank Carlson for her interest and thoughtful comments[1] regarding our exploratory study, "Assessing relatives' readiness for hereditary cancer cascade genetic testing."[2] We agree with Carlson that cascade genetic testing is an important but often challenging topic to study. Carlson notes several limitations of our study methods, acknowledged in our article, which we are happy to discuss in greater detail.

First, our decision to primarily recruit participants using social media and advocacy groups rather than a clinic-based approach was driven by our interest in working with a diverse and engaged population of individuals with hereditary predisposition to cancer, to replicate other successful hereditary cancer research recruitment approaches,[3] and to do so in a way that would be feasible and efficient given our study's resource constraints. Studies of cascade genetic testing have primarily recruited clinical patient populations, which also have limitations regarding representativeness. Carlson notes that our recruitment approach may have created a "highly motivated sample, unreflective of families that decline to engage";[1] however, our results demonstrate communication and cascade testing outcomes comparable to studies from primarily clinic-based populations. These results suggest that processes in families of social media–based patients may not be meaningfully different from families of clinic-based patients. The opportunity to leverage social media recruitment was a way to increase access for participants to engage in research; promote anonymity, thus decreasing barriers such as formal informed consent and medical record release authorizations; and to reach populations that may have otherwise been unable to participate via clinic-based recruitment.

We acknowledged in the limitations section that there were relevant data not collected in our survey including additional participant and relative characteristics (age, cancer history, etc.). As stated, anonymity and minimizing data entry burden on participants were important considerations when designing our study given the limited resources available to compensate participants for their time and effort, and in an effort to collect the minimum necessary data to answer our primary research question. We welcome opportunities for collaboration with Carlson and others to expand upon this initial exploratory work and to address these data gaps in future research.

We agree with Carlson that there are relevant limitations when considering information about relatives that are reported by study participants, which we also acknowledged in the article. Carlson specifically mentions the possibility of social desirability bias, which we agree can be present in any research study that relies on self-report whereby participants have an emotional investment or experience stigma. We reduced this bias in our study through anonymity of responses and lack of direct engagement with an interviewer or researcher when completing the electronic survey. We also agree that recall bias is relevant to all studies whereby a participant is asked to report on past events, and family communication (or lack thereof) may also be remembered or understood differently by relatives.[4] These potential sources of bias, relevant in studies of reported family communication and cascade genetic testing, are not unique to our study.

Finally, Carlson makes several points about the use of the transtheoretical model that we appreciate the opportunity to discuss. As was outlined in the discussion section of the article, there are many studies that have characterized determinants, barriers, and facilitators that exist within both the family communication and cascade genetic testing processes. Reassessing previously described factors known to influence cascade testing was not the aim of our study. We sought instead to explore the population of living, untested, at-risk relatives, particularly those aware of the hereditary cancer risk in the family, to see if there were different stages of cascade testing readiness present.

We argue that completing cascade genetic testing is a behavior, similar to other health behaviors like mammography, where an action can be observed. In contrast, a decision or intention to have testing does not necessarily cause or result in completed genetic testing. A strength of the transtheoretical model is that it is a theory of behavior change, unlike some other behavioral science theories that focus on prediction of possible future behavior.[5] We explained in our materials and methods section that in using the transtheoretical model as the framework for the study, the survey questions of readiness align with timing in the theory's stages of change construct. Carlson raised concerns about the utility of measuring stage of change, or readiness, of relatives in relation to the length of time since they first became aware of the genetic testing results in the family. Between initial awareness (precontemplation) and completion of the behavior (action), individuals may move between stages of change often in a nonlinear fashion. The amount of time any individual spends between precontemplation and ultimately reaching the action stage, if at all, is not predefined. Because our survey was only distributed once, we acknowledged that the stage of change distribution of relatives only represents one point in time; however, we view this baseline distribution of the living, at-risk, untested population as a starting point for further research.

A core assumption of the transtheoretical model is that no single model, including the transtheoretical model, accounts for all complexities of behavior change.[5] By using the model as a framework for our study we did not intend to suggest that this is the only explanation or theory for understanding the cascade testing process. We agree with the article Carlson cited by Armitage[6] in that components and uses of the transtheoretical model may be valuable especially using the stages of change for audience segmentation, and studying the processes of change, decisional balance, and self-efficacy. There are numerous opportunities for future research to improve our understanding of cascade testing decision-making and to use various behavioral science theories to inform interventions and programs to support families. We appreciate the opportunity to expand the discussion of our study, which we hope serves as one of many ongoing and future efforts to better understand how individuals move from initial awareness of hereditary cancer risk in the family to cascade genetic testing.

Erica M. Bednar[1,2]✉, Charlotte C. Sun[3], Sheryl McCurdy[4] and Sally W. Vernon[4]

[1]*Clinical Cancer Genetics, The University of Texas MD Anderson Cancer Center, Houston, TX, USA.* [2]*Cancer Prevention and Control Platform, The University of Texas MD Anderson Cancer Center, Houston, TX, USA.* [3]*Gynecologic Oncology and Reproductive Medicine, The University of Texas MD Anderson Cancer Center, Houston, TX, USA.* [4]*Department of Health Promotion and Behavioral Sciences, The University of Texas Health Science Center at Houston School of Public Health, Houston, TX, USA.*
✉*email: embednar@mdanderson.org*

Received: 18 December 2020; Accepted: 29 December 2020;
Published online: 23 March 2021

REFERENCES

1. Carlson, J. Correspondence on "Assessing relatives' readiness for hereditary cancer cascade genetic testing" by Bednar et. al. *Genet. Med.* (in press) (2020).
2. Bednar, E. M., Sun, C. C., McCurdy, S. & Vernon, S. W. Assessing relatives' readiness for hereditary cancer cascade genetic testing. *Genet. Med.* **22**, 719–726 (2019).
3. Rayes, N. et al. MAGENTA (making genetic testing accessible): a prospective randomized controlled trial comparing online genetic education and telephone genetic counseling for hereditary cancer genetic testing. *BMC Cancer* **19**, 648 (2019).
4. Himes, D. O. et al. Female family members lack understanding of indeterminate negative BRCA1/2 test results shared by probands. *J. Genet. Couns.* **28**, 950–961 (2019).
5. Prochaska, J. O., Redding, C. A. & Evers, K. E. The transtheoretical model and stages of change. In *Health Behavior: Theory, Research, and Practice* (eds. Glanz, B., Rimer, B. K. & Viswanath, K.) 125 (Jossey-Bass, San Francisco, 2015).
6. Armitage, C. J. Is there utility in the transtheoretical model? *Br. J. Health Psychol.* **14**, 195–210 (2009).

COMPETING INTERESTS

The authors declare no competing interests.

ADDITIONAL INFORMATION

Correspondence and requests for materials should be addressed to E.M.B.

Reprints and permission information is available at http://www.nature.com/reprints

Publisher's note Springer Nature remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.