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Introduction to the Special Issue on Clinical and Public Health Genomics: Opportunities for translational behavioral medicine research, practice, and policy

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Opinions, and the current evidence base, vary widely about whether and how genetic and genomic (herein "genomic") information can be used to improve the public's health [1–3]. In the rapidly changing contexts of clinical care and genomic science discovery, a growing body of translational research has emerged to evaluate, integrate, and apply genomic risk information for improved individual and collective health outcomes. In the current issue of *Translational Behavioral Medicine*, we highlight research evaluating the use of genomic information across the healthcare spectrum and within primary, secondary, and tertiary levels of disease prevention and control.

The first set of articles report new findings largely within the context of primary prevention and include a critical commentary that raises and addresses skepticism over genomic applications in behavioral medicine. Using an evaluation process that could be applied across various genomic discoveries, Ramsey et al. [4] reviewed the science related to variation in the alpha 5 nicotinic cholinergic receptor subunit gene *CHRNA5* and smoking outcomes (behaviors and related diseases). The authors posit that genomic testing can inform disease risk, optimize smoking cessation treatment, and motivate smoking behavior change—although future research is needed to focus on establishing clinical utility [4]. Complementing the review by Ramsey et al. [4], the work of Wright et al. [5] describe outcomes from a qualitative study of participants in a randomized, controlled trial examining perceptions of the use of genomically informed treatment dosing in a multi-modality behavioral and pharmacologic cessation intervention. The authors report that relative to the perceived importance of the primary intervention (i.e., dual behavioral support coupled with nicotine replacement therapy), the perceived importance of pharmacogenomic tailoring of nicotine replacement therapy was comparably modest and unassociated with strengthened adherence. Within a different primary prevention context, Aspinwall et al. [6] describe the relevance of genomic predisposition to melanoma as represented by the identification of a germline mutation in the *CDKN2A* (p16) gene. Compared with individuals who received family health history information, those who received family health history information plus results from *CDKN2A* genomic testing reported feeling more informed about their cancer risks and expressed stronger motivations to reduce their sun exposure (a major risk factor for melanoma). However, mutation carriers who were parents reported more worries over their children's risk than those who did not undergo testing. This raises the need for translational behavioral research to better inform preventive health efforts when the focus is on children and genomics.

Children and genomics are the topic of the work by Kulchak-Rahm et al. [7] who explored responses of parents to two types of genomic information about their children: (1) a medically treatable childhood disease and (2) an adult-onset hereditary cancer syndrome. Not surprisingly, parents were motivated to learn the results about their healthy children in an effort to help prepare them for their future healthcare needs. In a provocative commentary by McBride [8], the author reviews and rebuts three recurrent critiques of genomics and its integration into behavioral and public. Through this engaging discussion, examples of research highlighting the benefits and significant potential of genomics are presented, supporting the value of investing time and resources into behavioral medicine and public health research on this topic.

The second set of articles explores elements of the processes and contents by which genomics can improve outcomes among those at increased risk for/already affected by cancer and chronic diseases. As cardiovascular disease and cancer continue to be leading preventable causes of morbidity and mortality in the United States [9], they do remain a major focus of prevention efforts among translational behavioral medicine researchers, clinicians, and policy makers, including those incorporating genomic perspectives in their work. Five articles in this section of this issue are emblematic of that perspective. First, the work of Peterson et al. [10] examined published research on health communication

surrounding cancer genomic testing. In it, they chronicle nearly a decade's worth of progress in the field, including the importance of basic behavioral research on patient-provider communication and family communication, to improve the uptake and outcomes of testing. Moreover, they note persistent disparities in the translation of genomic findings to underserved groups and communities—an important topic that remains a focus for the field at large [11]. Similarly, the work by Kaphingst et al. [12] and a related commentary by Vadaparampil and Cragun [13] address issues confronting young patients with breast cancer attempting to access information from advanced genomic sequencing, highlighting the important role of shared decision making, psychosocial, and cultural factors, and system-level issues that affect the availability and accessibility of this information. In much the same way, Ricker et al. [14] report on the results of a prospective investigation of family communication of at-risk genomic status in a racially and ethnically diverse study sample. They found the prevalence of family communication to be high, with nearly one-third of potentially at-risk relatives undergoing genomic testing themselves. In an era of precision medicine, effort to reach relatives unaffected by disease remains an aspirational goal to prevent its further spread within families. Finally, the paper by Skapinsky et al. [15] remind us of the importance of kinship networks in behavioral and preventive medicine research, and ways to tap their potential as change-agents within family systems.

The third section in this special issue underscores approaches to integrating genomic information into clinical care and behavioral medicine research, with attention to cross-cutting themes. The works highlight areas important to health care and translational behavioral medicine research: communication and interpretation of genomic results, informed consent and relevant stakeholder perspectives, and behavioral responses to different genomic testing contexts. Allen et al. [16] deployed mixed-method approaches to evaluate the characteristics of patients who purchase third-party genomic testing services and the experiences of genetic counselors in interpreting and educating patients about their results. Genetic counselors reported a number of challenges, including patients' overemphasis on the validity of the genomic data and resistance to information provided by the counselor. The growing availability of third-party genomic testing companies necessitates future attempts to support patients and healthcare professionals in these evolving contexts [16]. The influence of variable contexts and perspectives about genomics was further evaluated in the work by Goodman et al. [17] that examined views from three stakeholder groups about the use of de-identified genomic data. Research participants, genomic researchers, and Institutional Review Board (IRB) professionals held differing perspectives, with researchers reporting less concern than participants or IRB professionals about the possibility that participants could be personally identified or harmed from a study involving de-identified data. Expanding on participant perspectives, a second work by Goodman et al. [18] conducted a systematic review to identify participant views related to consent, use of de-identified genomic data, and return of individual genomic results. While most participants supported the return of individual genomic results and broad informed consent, differences in views about consent were evident across participant age and gender. Participant responses to genomic information can also be shaped by financial variables as identified by Lui et al. [19] in an innovative study that examined the influence of co-payment on behavior. Using results from a longitudinal cohort and an online experiment, when individuals had to pay out-of-pocket costs to receive genomic test results they were more likely to share these results with their physicians and obtain health screenings. Bridging these areas of clinical application and research, Koehly et al. [20] provide a commentary about the essential role that clinical and behavioral sciences play in genomic translation. The authors highlight priority areas surrounding the integration of behavioral, social, and neurocognitive factors, informed translation of genomic discoveries, and the need for increased outreach and education efforts at multiple levels. The commentary concludes with examples of current and future federal intramural and extramural research programs related to these high-priority areas.

We trust that the type and variety of perspectives portrayed in this special issue —spanning patient perspectives and health outcomes, clinical contexts and implications for policy and research — ignite readers' interest, engagement, and investment in clinical and public health genomics. The accumulating evidence is promising, with clear examples spanning the continuum from the earliest stages of translation to clinically important applications that improve public health [21]. The current efforts, and resulting evidence, are a critical part of the "discovery to delivery" research pipeline. The articles here highlight the potential of including behavioral medicine practitioners and scientists "at the table" with genomic researchers, payers, and funders to better understand, interpret, and use such data for maximal benefit.

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References

1. Khoury MJ, Galea S. Will Precision medicine improve population health? *JAMA.* 2016;316(13):1357–1358.
2. Phillips KA, Deverka PA, Sox HC, et al. Making genomic medicine evidence-based and patient-centered: a structured review and landscape analysis of comparative effectiveness research. *Genet Med.* 2017;19(10):1081–1091.
3. Roberts MC, Kennedy AE, Chambers DA, Khoury MJ. The current state of implementation science in genomic medicine: Opportunities for improvement. *Genet Med.* 2017;19(8):858–863.
4. Ramsey AT, Chen L, Hartz SM, et al. Toward the implementation of genomic applications for smoking cessation and smoking-related diseases. *Transl Behav Med.* 2018;8:7–17.
5. Wright AJ, Sutton S, Armstrong D, Aveyard P, Kinmonth AL, Marteau TM. Factors influencing the impact of pharmacogenomic prescribing on adherence to nicotine replacement therapy: A qualitative study of participants from a randomized controlled trial. *Transl Behav Med.* 2018;8:18–28.
6. Aspinwall LG, Stump TK, Taber JM, et al. Genetic test reporting of CDKN2A provides informational and motivational benefits for managing melanoma risk. *Transl Behav Med.* 2018;8:29–43.
7. Kulchak-Rahm A, Bailey L, Fultz K, et al. Parental attitudes and expectations toward receiving genomic test results in healthy children. *Transl Behav Med.* 2018;8:44–53,
8. McBride CM. Translation research to apply genomics to improve health promotion: Is it worth the investment? *Transl Behav Med.* 2018;8:54–58.
9. García MC, Bastian B, Rossen LM, et al. Potentially preventable deaths among the five leading causes of death – United States, 2010 and 2014. *mmwr Morb Mortal Wkly Rep.* 2016;65(45):1245–1255.
10. Peterson EB, Chou WS, Gaysynsky A, et al. Communication of cancer-related geneic and genomic information: A landscape analysis of reviews. *Transl Behav Med.* 2018;8:59–70.
11. Graves KD, Tercyak KP. Introduction to the special series on health disparities in genomic medicine. *Health Psychol.* 2015;34(2):97–100.
12. Kaphingst K, Ivanovich J, Lyons S, et al. Preferences for learning different types of genome sequencing results among young breast cancer patients: Role of psychological and clinical factors? *Transl Behav Med.* 2018;8:71–79.
13. Vadaparampil ST, Cragun D. Shared decision making: Implications for return of results from whole exome and whole genome sequencing. *Transl Behav Med.* 2018;8:80–84.
14. Ricker CN, Koff RB, Qu C, et al. Patient communication of cancer genetic test results in a diverse population. *Transl Behav Med.* 2018;8:85–94.
15. Skapinsky KF, Persky S, Lewis M, et al. Heart disease risk information, encouragement, and physical activity among Mexican-origin couples: self- or spouse-driven change? *Transl Behav Med.* 2018;8:95–104.
16. Allen C, Gabriel J, Flynn M, Cunningham TN, Wang C. The impact of raw DNA availability and corresponding online interpretation services: A mixed methods study. *Transl Behav Med.* 2018;8:105–112.
17. Goodman D, Johnson CO, Bowen D, Smith M, Wenzel L, Edwards KL. A comparison of views regarding the use of de-identified data. *Transl Behav Med.* 2018;8:8:113–118.
18. Goodman D, Bowen D, Wenzel L, et al. The research participant perspective related to the conduct of genomic cohort studies: A systematic review of the quantitative literature. *Transl Behav Med.* 2018;8:119–129.
19. Liu W, Outlaw JJ, Wineinger N, Boeldt D, Bloss CS. Effect of co-payment on behavioral response to consumer genomic testing. *Transl Behav Med.* 2018;8:130–136.
20. Koehly LM, Persky S, Spotts E, Acca G. Social and behavioral sciences research priorities for genomic translation. *Transl Behav Med.* 2018;8:137–143.
21. Khoury MJ. No shortcuts on the long road to evidence-based genomic medicine. *JAMA.* 2017;318(1):27–28.