Genomic medicine can revolutionize the personalization of disease risk and optimize the adoption of clinical and behavioral recommendations designed to promote health (Collins, Green, Guttmancher, & Guyer, 2003). An examination of such approaches may lead to behavior change through precision prevention (Khoury & Evans, 2015). Despite this promise, genetic testing—whether offered within a clinical context or directly to consumers—has shown limited reach and inequitable access (Green & Guyer, 2011; Weitzel, Blazer, MacDonald, Culver, & Offit, 2011).

According to Healthy People, 2020, there are several looming challenges in genetics, including: “Monitoring the use of genetic tests and family health history in populations, the health outcomes related to their use, and disparities in their use and outcomes” (Healthy People 2020, 2017). Beneficiaries of learning personalized genetic information have traditionally been non-Hispanic White individuals at higher socioeconomic and education levels, with insurance coverage, and who have higher health literacy (Bloss, Schork, & Topol, 2011). Members of racial and ethnic minority groups and those with lower education levels also stand to benefit from genomics, but are less likely to participate in genomic research or undergo genetic testing when offered (Bloss et al., 2011; Hensley Alford et al., 2011; Suther & Kiros, 2009). If genomic medicine is to make a significant difference in behavior change, and have a broader impact on precision prevention and healthcare delivery, it must reach individuals who are known to have limited healthcare access (Kaphingst & Goodman, 2016).

Systematic evidence reviews have examined the effects of genetic risk feedback (e.g., from use of genetic tests or family health histories) on changes in health behaviors across domains including diet, physical activity, smoking cessation, and alcohol use (Stewart, Wesselius, Schreurs, Schols, & Zeegers, 2017). These reviews concluded that interventions providing personalized genomic information are not generally effective at promoting behavior change. However, the studies reviewed have been largely comprised of White, highly educated participants. In one review (Hollands et al., 2016), only 3 of 18 studies included diverse samples. As such, these findings do not specifically examine the role of sociodemographic characteristics such as race, education, or literacy that may be informative for understanding the implications of genetic risk information for health behaviors in diverse populations.

In parallel, a recent systematic review of literature examining translational opportunities (utility and actual use of genetic technologies, rather than basic gene identification) identified substantial deficits in research involving diverse populations (Roberts, Kennedy, Chambers, & Khoury, 2017). Using the Centers for Disease Control and Prevention’s Public Health Genomics Knowledge Base,
the authors found that diverse populations have been underrepresented in research efforts moving basic genomics findings into clinical and community settings. Nearly half (44%) of the 283 reviewed articles were missing racial/ethnic breakdown data. Of the articles reporting the racial/ethnic composition of the sample, most (77%) consisted exclusively of Whites. Only 24% of these studies included Blacks, 16% included Asians, and 13% included Hispanics. Most of these studies were conducted in clinical (65%) rather than public health settings (24%), including community-based studies or population-based registries, further limiting generalizability to the wider population. This lack of participant diversity is a prominent limitation of this literature, and a strong rationale not to foreclose research examining behavior change opportunities in translational genomics before further work can be done to broaden the reach of studies designed to evaluate this potential (Graves, Hay, & O’Neill, 2014; McBride, Birmingham, & Kinney, 2015).

This homogeneity among study participants could be attributed to the fact that genetic testing has primarily only been available in high-risk clinics located largely in cancer centers and other specialty clinics. Yet, even with the advent of accessible direct-to-consumer (DTC) genetic testing, study participants and DTC genetic test users are—as in the behavior change research literature—most frequently White, well-educated, and insured (Bloss et al., 2011; Stewart et al., 2017). With the widening availability of genomic technologies, and the increasing knowledge base concerning genomics and health outcomes, understanding how diverse participant characteristics may influence effects of genetic risk feedback on motivation for behavior change is becoming increasingly important (Li, Ye, Whelan, & Truby, 2016; Marteau et al., 2010). In the next sections, we review examples of ongoing efforts to understand genetics and behavior across diverse populations.

**Genomics and Behavior in Diverse Populations**

**Smoking Behavior**

Several studies in the area of behavior and genomics have examined genetic testing for susceptibility to lung cancer and nicotine addiction among African Americans, where death from smoking-related disease is disproportionately high (Centers for Disease Control and Prevention, 2017). An early study (McBride et al., 2002) recruited low-income African American smokers to undergo a biomarker feedback (BF) intervention or enhanced usual care for smoking cessation. The BF intervention offered a blood test for the \( \text{GSTM1} \) gene, which is associated with susceptibility to lung cancer, and 83% of participants assigned to this group agreed to be tested. At six months, those in the BF group had significantly higher rates of smoking cessation, but this difference did not persist at 12 months, which may indicate the need for strategies to enhance behavioral maintenance. Within the BF group, cessation rates did not differ by actual risk feedback. One limitation of this work was that 45% of the smokers did not fully understand their test results. Lipkus and colleagues (Lipkus, McBride, Pollak, Lyna, & Bepler, 2004) further examined data from this trial to evaluate how participants interpreted the \( \text{GSTM1} \) feedback. Participants who received higher genetic risk feedback were less likely to accurately recall their result than those who received average genetic risk feedback. Education levels and baseline risk perceptions were important correlates of accuracy of recall. Those with lower perceptions of risk at baseline and with lower education were more likely to inaccurately recall and to misinterpret their findings.

These studies highlight the importance of improving the effects of genetic risk information interventions over time and enhancing clarity in genetic risk feedback. Ideally, studies should perform short-term follow up to confirm comprehensibility of feedback, as well as long-term follow up to assess maintenance of behavioral outcomes. Another limitation of this trial was that most participants were ready or attempting to quit smoking, and were thus already highly motivated, independent of...
intervention condition. Reaching individuals who are less motivated is an important strategy for understanding the process behind behavior change, and recruiting participants with a wide range of motivation will prevent ceiling effects.

A recent study examined behavioral and psychosocial reactions to genetic testing for risk for common tobacco-related diseases (e.g., diabetes, heart disease, cancers) in a sample of nicotine-dependent smokers, with limited health literacy, of whom 64% were African American (Hartz et al., 2015). Most (61%) were interested in receiving their test findings. Importantly, quit attempts increased from 21% to 53% at eight-week follow-up, all but one participant discussed test findings with physicians or family and friends, and distress symptoms were not increased with testing or by test result. This study suggests that there may be potential to deliver smoking cessation interventions after the receipt of genetic testing results, as those tested may be primed and more highly motivated for cessation after receiving their results.

Additional research on receptivity to testing, rather than behavioral outcomes per se, indicates that African Americans might be relatively receptive. For instance, among African American college students who had experimented with smoking or had favorable smoking attitudes, interest in genetic testing for lung cancer risk was high overall, but highest among those who believed themselves to be at high risk and who believed lung cancer was influenced by genetics (McBride, Lipkus, Jolly, & Lyna, 2005). In a study of interest and intentions to participate in genetic risk research among adult African American smokers (Halbert, Gandy, Collier, & Shaker, 2006), most (58%) reported they were likely to take a genetic test to identify risk factors for lung cancer. Beliefs about the benefits and risks of participating in medical research and genetic testing predicted receptivity to genetic testing. Other studies (Park et al., 2011) show similar percentages of African Americans reported likeliness to participate in genetic testing for nicotine addiction (62%). However, 91% of White participants in the sample indicated likeliness to test, signifying some reluctance to consider testing among African Americans, at least in comparison to White participants. The study identified barriers among African Americans that may explain this finding, including disinterest in testing, skepticism, and the belief that genetics do not play a role in this behavior. This research supports previous findings that African Americans are less likely to complete genetic testing due to potential mistrust of the medical system (Peters, Rose, & Armstrong, 2004). Protocols that address these concerns and discuss the benefits and limitations of genetic testing may facilitate higher interest and willingness to test among African Americans (Halbert et al., 2006).

**Energy Balance**

One study of energy balance-related behaviors provided genetic risk information about type II diabetes mellitus (DM), and recruited a racially diverse sample that was 53% African American (Voils et al., 2015). Most (80%) participants were male, and all were obese/overweight but not diabetic. The intervention group received personalized genetic risk information for DM, while the control group received counseling about eye disease. All participants were encouraged to lose weight to prevent DM onset. Calorie and fat intake were lower in the intervention group at three but not six months. Insulin resistance, perceived risk, and physical activity did not differ at either time point. There were no differences in outcomes within the intervention group by actual risk feedback. Intensive, long-term interventions are the gold standard for weight loss, and future studies should examine whether genetic risk information can be incorporated to enhance DM prevention intervention delivery. As with smoking cessation, the question of how to maintain longer term behavior change exists, along with the question of whether level (higher/lower risk) or just the receipt of results based on genetic testing compared to non-genetic test results prompts behavior change.
Genomics and Behavior Change

Alcohol Use

One study (Hendershot, Otto, Collins, Liang, & Wall, 2010) investigated the effect of genetic risk feedback in motivating reductions in alcohol consumption in a college-based sample of Asian Americans. The ALDH2*2 allele is rarely found outside of those of northeast Asian descent, appearing in approximately 540 million individuals worldwide. While ALDH2*2/*2 genotype appears to protect against alcohol abuse and dependence due to physiological reactions to alcohol consumption, those with the ALDH2*1/*2 genotype are at risk for moderate to heavy drinking, and are at elevated risk for alcohol-related cancers (compared to those with ALDH2*1/*1 genotype who face the highest risk for alcohol dependence). In a randomized trial, participants in the intervention group were provided genetic feedback and risk information about alcohol-related cancer or alcohol dependence (dependent on genotype). Significantly reduced drinking frequency and quantity resulted among participants with the ALDH2*1/*2 genotype after 30 days. Additionally, participants in the intervention group demonstrated significantly increased fear arousal, intentions to change drinking behavior, and risk perceptions. Overall acceptability of the intervention was high, measured by questions about how interesting, useful, informative, and engaged participants were with the genetic feedback.

Cancer Screening

A recent trial (Weinberg et al., 2014) examined outcomes of genetic and environmental risk assessment for moderate increases in risk of colorectal cancer among diverse participants recruited from hospital-based primary care clinics (35% African American, 26% had high school education or less). No differences in colorectal cancer screening adherence were observed between the genetic risk feedback and control groups. However, a secondary analysis of the trial data (Myers et al., 2015) found that among African American participants, elevated genetic risk feedback was more likely to lead to higher anxiety, whereas among White participants, elevated genetic risk feedback was more likely to prompt colorectal cancer screening (screening occurred in 67% of Whites vs. 33% of African Americans). These differences were not observed in the average risk groups. Focus groups revealed that many participants believed that genetic testing was a replacement for screening tests. African American participants experienced strong negative reactions to elevated genetic risk feedback and explained how this discouraged them from screening due to fear of being “too late.” These findings highlight some distinct behavioral and psychosocial reactions to genetic risk information that should be replicated across behavioral contexts, and dictate a need for tailored information for those who are highly distressed, and addresses perceptions of diverse populations. An additional study (Graves et al., 2013) of genomic testing for colorectal cancer risk in a university-based internal medicine clinic (43% non-White, most participants adherent to screening at baseline), found high levels of improvement in exercise and eating behaviors in those tested after three months, although behavioral outcomes did not differ by risk status. African American participants were more likely than White participants to share their results with their physicians.

Research conducted in the high-risk setting examined behavioral outcomes of genetic counseling and testing for BRCA1 mutations in an African American kindred, 90% of whom identified as Creole ethnicity (Kinney et al., 2006). Among 10 BRCA1 mutation carriers assessed one year following genetic testing, participants were more likely to pursue enhanced breast cancer screening than prophylactic mastectomy. The authors propose that this might be related to specific values and attitudes in this population (e.g., surrounding body image, risk reduction options, and patient-provider communication), and differences in insurance reimbursement and other access factors. Perhaps more importantly, even though rates of mammography screening improved in mutation carriers, there was a high level of non-adherence to mammography at baseline that was not resolved at one-year
follow-up. These findings indicate a need for further culturally sensitive research with this unique population.

**Future Research Directions**

In 2015, the National Academy of Sciences, Engineering and Medicine convened experts in the field of genomics for a workshop on applying an implementation science approach to genomic medicine. The group identified the topic of disparities as of critical importance. Members stated: “In order to ensure equitable access to genomic medicine, greater efforts will be required to address health inequities across low income and minority groups. [...] Genomics will only achieve its full potential to improve health when the advances it engenders become accessible to all” (National Academy of Sciences Engineering and Medicine, 2016). Next we review some important rate-limiting factors in moving this field forward, and then provide an example of a national effort that may start to address these limitations.

**Access**

Understanding behavioral outcomes rests on adequate access to genomic technologies. There is an emerging research base regarding genetic attitudes and preferences in underrepresented communities. In general, minority populations have been interested in genetic information but are less likely to access it (Hamilton et al., 2016; Kaphingst et al., 2015). Members of underserved populations report great interest in genomics especially when information is presented in an accessible and relevant way to their sociocultural and behavioral characteristics (Kaphingst et al., 2015; Sussner et al., 2011; Torres et al., 2014). In a study examining interest in multiplex genetic testing for risk for eight common health conditions including cancer and heart disease in diverse primary care patients in Detroit, Michigan, about 30% expressed interest in testing, yet race remained an important predictor of interest with Whites showing more interest than Blacks (Hensley Alford et al., 2011; McBride et al., 2009).

Translating genetic information to diverse populations raises significant but addressable challenges. Racial and ethnic minority groups vary in their understanding and linguistic elaboration of genetic concepts (Kaphingst & Goodman, 2016). As a result, there is a great need for the development of low literacy, multilingual genomic risk education materials. Moving from a high resource, time intensive genetic counseling approach to greater reliance on more generalizable channels for information dissemination (e.g., the Internet), may be a promising vehicle to make genomic information more accessible. The potential to minimize widening health disparities is great; the challenge is to develop, confirm, and validate such efforts with detailed input from the target population to propel the field forward.

**Theoretical Frameworks**

In studies examining the role of genetics in promoting health behavior change, individual-level factors (e.g., awareness, knowledge, attitudes, beliefs) have been heavily examined, yet system-level factors (e.g., health insurance, misuse of information, discrimination, mistrust) will also be important for understanding access issues that may contribute to disparities, and act as modifiers of behavior change (Kaphingst & Goodman, 2016). For example, recent research with Hispanics suggests that when barriers to testing are removed (i.e., access, cost), a large proportion are very likely to pursue genetic risk assessment and testing and are also willing to share bio-specimens for research purposes (Komenaka et al., 2016; Nodora et al., 2017). Consequently, multi-level theoretical models (Taplin
et al., 2012) will be necessary to develop and evaluate culturally relevant outreach approaches that address individual and systemic barriers to genomic medicine. Additionally, implementation science frameworks, such as Diffusion of Innovations Theory (Rogers, 1995) and RE-AIM (Glasgow, Vogt, & Boles, 1999), have been rarely used in translational genomics research (Roberts et al., 2017) and could help address disparities relevant to behavior and genomics.

**Trust**

While systemic influences such as access and socioeconomic status curtail inclusivity in genetics research, some studies demonstrate that mistrust is another critical barrier. One qualitative study (Scharff et al., 2010) found that mistrust was identified as an important barrier to research participation in general by African Americans in 11 focus groups. Participants cited historical violations of trust such as the Tuskegee syphilis study, as well as current systematic discrimination. A study that controlled for education, income, and other demographic variables in multiple cohorts showed African American and Hispanic participants are more likely than Whites to believe that racial minorities are more likely to be taken advantage of in biomedical studies than are Whites (Katz et al., 2008). Regarding predictive genetic testing specifically, concerns reported by African American and Latino participants include that the government may use genetic tests to label groups as inferior (Peters et al., 2004), and that genetic information may generally be misused (Suther & Kiros, 2009; Thompson, Valdimarsdottir, Jandorf, & Redd, 2003). Better outreach and trust-building are needed. One important step toward this goal was the passage of the Genetic Information Nondiscrimination Act (GINA) in 2008, which was designed to protect Americans against discrimination based on genetic information (Saulsberry & Terry, 2013).

**All of Us**

A national effort is gaining momentum to enroll large segments of the general population for research on genomic, behavioral, and environmental exposure factors to galvanize research in precision prevention (National Institutes of Health, 2017). The effort is designed to be a research resource to address a variety of diseases and health questions, and will be particularly focused on diverse populations, such as those living in remote areas, people with multiple chronic conditions, sexual and gender minorities, and those with lower educational attainment or income. Importantly, targeted outreach materials will be developed to serve these populations. This effort seeks to go well beyond prior attempts at inclusion to break down both attitudinal and access barriers to achieve equitable representation within the program. This outreach will be especially valuable for engaging highly mobile populations such as migrant workers, shelter populations, and those in temporary housing. Efforts will leverage existing community network relationships and use planned retention techniques (e.g., reminder calls, outreach calls to an authorized relative/friend, home visits). This research effort will also create mechanisms for the return of relevant research results to participants, such as results reflecting genetic risk information.

**Conclusions**

Given the limits of past research, there are multiple opportunities for broadening and deepening our understanding of how genomic information may influence a range of behaviors in diverse populations. Latinos, in particular, have received relatively little focus in this body of research. Research engaging Latinos will need to address issues of health literacy, language, and access (Hamilton et al., 2016), as well as cultural values such as the importance of familial relationships (Ashida, Wilkinson, &
Koehly, 2012). Diverse US sub-populations may be more or less engaged by genetic risk feedback and behavior change, and require study. There is also a need to develop novel strategies to communicate genetic test results in a way that promotes high comprehension. Information tailoring may be appropriate across various languages, ages, and ethnicities. In one effort to motivate sun protection and skin cancer screening using genetic risk feedback in Hispanics utilizing primary care, the authors conducted Spanish translations and cognitive interviews to confirm comprehensibility among New Mexico Spanish speakers (Hay et al., 2017; Rodriguez et al., 2017).

There is also a need for more theory-informed research to examine whether factors such as illness fatalism, medical mistrust, or perceived control over health behaviors are important moderators of effects that may be common (or not) across diverse populations. In a related vein, further research examining individuals’ beliefs about the underlying causes of diseases will help shape future interventions. Research indicates that multifactorial causal beliefs about health conditions are fairly common in the US (Waters, Muff, & Hamilton, 2014), and may be associated with sociodemographic characteristics. Culturally specific causal beliefs about illness are important elements as well (Palmquist, Wilkinson, Sandoval, & Koehly, 2012).

Despite the inconsistent evidence to date that genetic risk feedback motivates behavior change, we assert that research is needed to clarify what types of intervention components (dictated by theory and other behavior change research outside of the context of genetics) should be added to genetic risk information to maximize motivational potential and to examine this across diverse populations. Indeed, longer term behavior change may require booster sessions and ongoing support as well as consideration of unique factors relevant to specific populations. Given continuing advances in genomics technology, as well as efforts such as All of Us that are designed to engage diverse populations, in the coming years genetic susceptibility information will be more relevant to the general population, and likely even more available via DTC testing and traditional primary care outlets. Undoubtedly, our national investment in genomics research should benefit us all. Inclusive research strategies instituted in the near-term could help narrow these gaps in the coming years.

References


Genomics and Behavior Change
Jennifer L. Hay, Jennifer M. Bowers, and Jada G. Hamilton


Genomics and Behavior Change

