Addressing underrepresentation in genomics research through community engagement

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Summary

The vision of the American Society of Human Genetics (ASHG) is that people everywhere will realize the benefits of human genetics and genomics. Implicit in that vision is the importance of ensuring that the benefits of human genetics and genomics research are realized in ways that minimize harms and maximize benefits, a goal that can only be achieved through focused efforts to address health inequities and increase the representation of underrepresented communities in genetics and genomics research. This guidance is intended to advance community engagement as an approach that can be used across the research lifecycle. Community engagement uniquely offers researchers in human genetics and genomics an opportunity to pursue that vision successfully, including by addressing underrepresentation in genomics research.

Introduction

The American Society of Human Genetics (ASHG) is committed to creating a future in which all people are able to realize the benefits of human genetics and genomics research (https://www.ashg.org/about/mission-strategic-plan/). Such a future holds promise: initiatives in precision medicine are creating the opportunity to improve care for both healthy individuals and those with various medical conditions. The commitment to make these benefits relevant to *all* people will require focused efforts to address health inequities and remove barriers to increase representation of diverse communities in genetics and genomics research. Addressing underrepresentation, however, is not merely a matter of adapting recruitment efforts.

Factors such as systemic racism, historical abuses, structural discrimination, and lack of knowledge on the benefits of research participation might preclude underrepresented groups from engaging in research. Researchers, healthcare providers who contribute to research recruitment, and research institutions might also inadvertently perpetuate these structural barriers to participation through implicit or unconscious biases, as well as the development of research infrastructure and practices that fail to promote broader engagement or inclusion. For this reason, efforts to broaden benefits must begin with meaningful engagement with historically and currently underrepresented communities.

Contemporary genomics research involves collaboration across a wide spectrum of basic scientists, translational researchers, clinical trialists, outcomes researchers, and ethical, legal, and social implications (ELSI) scholars as

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Box 1. Key terms and definitions

Benefit sharing: the action of giving a portion of gains, profits, or other benefits derived from the use of human genetic resources to the resource providers in order to promote justice in exchange. Benefit sharing often emphasizes the clear provision of benefits to those who might lack reasonable access to anything of value, especially health-related, resulting from the research.[4–6]

Community: groups of people who are affiliated by geographic proximity, shared interests, history, culture, and tradition or similar situations and experiences with respect to issues affecting their well-being.[2] Heterogeneity within communities is universal, and lack of external recognition does not detract from a group of people's being a community.

Cultural humility: a lifelong commitment to self-evaluation and critique ; reflexivity on one's own implicit or unconscious biases; addressing power imbalances; and developing mutually beneficial and non-paternalistic partnerships with communities on behalf of individuals and defined populations.[7,8]

Ethnicity: a subjective, socially constructed concept employed to refer to groups of individuals who share a similar cultural heritage or identity (history, language, and/or religion), where these characteristics have social meaning.[9,10]

Genetic ancestry: refers to inferences about ancestral origin, an individual's lineage of descent, or the geographic history of an individual's ancestors according to signatures in their DNA.[11]

Group harms: damages or injury, tangibly experienced or perceived, that impact the welfare interests of a group and its people.[12–14]

Race: a subjective, dynamic, and complex social construct, generally employed to group individuals on the basis of observed (or ascribed) biological or phenotypic traits, where these characteristics have acquired socially significant meaning.[9–11,15,16]

Stakeholder: an individual, group of individuals, or organization that has a vested interest or a stake that can affect or be affected by a course of action.[17]

Structural discrimination: the pervasive embedded advantage and disadvantage within systems and other structures, including but not limited to political, legal, economic, medical, and research arenas, that produce, condone, and perpetuate widespread unfair treatment to minoritized groups, including LGBTQIA+ people, people with disabilities, and others.

Systemic racism: the pervasive embedded advantage and disadvantage within systems, including but not limited to political, legal, economic, medical, and research arenas, that produce, condone, and perpetuate widespread unfair treatment of people of color.[1,18]

well as patient advocates and health inequity scholars.[1] Given the collaborative nature of this work, all ASHG members need to be familiar with the importance of community engagement and take responsibility for these efforts in ways that are appropriate to their role. Fundamentally, establishing the trustworthiness of research projects begins with building mutually respectful long-term relationships, working towards adopting trustworthy practices, and broadening participant representation in research. This requires collaboration and commitment across the full spectrum of genetics and genomics stakeholders. This commitment recognizes that the participation of diverse groups is only the initial step towards broader representation across the full research cycle, including input into research questions and prioritization, processes for return of results, and the translation of studies towards clinical benefit.

This guidance document is focused on providing genetics and genomics researchers with a starting point for building trustworthy relationships and working with communities who are historically marginalized and underrepresented in genetics and genomics research. Although the specific strategies utilized will vary depending on a variety of factors, such as community needs and preferences, study aims, and funding availability, the principles of community-engaged research provide a critical methodological starting point.[2,3] This guidance describes the fundamentals that are shared across approaches for community-engaged research, as well as practical guidance on how to approach research with meaningful community interactions. Box 1 provides some provisional definitions of terms that are key to this discussion, although we acknowlege that the meanings of many of these terms remain the subject of active discourse and debate.

The representation problem

Genetics and genomics research focused on human health is fundamentally dependent on leveraging genetic diversity to identify genetic variants that contribute to disease risk or resilience, enabling diagnosis and prognosis. It drives development of novel therapeutics, indicates appropriate application of precision therapies, and predicts individual therapeutic responses. Inadequate representation of people of non-European ancestry groups leads to an incomplete understanding of human genetic diversity. However, persons do not generally identify on the basis of their ancestry but rather affiliate with communities on the basis of geographic proximity, shared interests, history, culture, and tradition. Therefore, representation of diverse communities is important for bringing the benefits of genetics and genomics to all and is also an important strategy for addressing underrepresentation of non-European ancestry groups.

The long-standing history of worldwide systematized oppression, discrimination, and colonization has created a contemporary world in which individuals of non-European ancestry often also identify with communities that have been exposed to oppression and exclusion. In the research realm, including the field of human genetics and genomics, instances of oppression and exclusion such as research abuses and eugenic sterilization have led to mistrust among populations that have been,[19–22] and continue to be, marginalized and underrepresented in research.[23,24]

Underrepresentation is problematic from both a scientific and an ethical point of view. It decreases our knowledge of genetic diversity across populations, thus failing to capture the full spectrum of genetic variation, and limits the global applications for prognosis, disease prevention, and therapeutic identification. More importantly, this can affect the transferability of research findings and practical applications to improve health outcomes. Underrepresentation also has a disproportionately adverse impact on the groups who are already marginalized, potentially leading to fewer actionable insights for precision medicine interventions in these populations and thus exacerbating health disparities. As a result, individuals of non-European ancestry who are underrepresented in the underlying research might disproportionately receive little benefit from it. Therefore, it is critically important to address these disparities in future genomics research to avoid generation of inaccurate scientific knowledge, inequitable distribution of the benefits of precision medicine, exacerbation of existing health disparities and other inequities created by systemic racism, stigmatization, biases in interpretation, and inattention to related social determinants of health.

If the genetics and genomics community is to avoid perpetuating these inequities, researchers must start by earning the trust of underrepresented communities and adopting practices that demonstrate trustworthiness. Given the long history of systemic racism and structural discrimination ingrained within the research enterprise, earning trust will require addressing biases, discrimination, and stigmatization at each level of research design, fair and equitable implementation of the research, careful attention to benefit sharing, and communication of findings. Researchers can begin by finding out from individual communities how genetics and genomics research might address the community's priorities and needs. Investigators should invite and empower communities to become partners in the research process, to collaboratively shape the research to reap its benefits, identify barriers, offer accommodations to make participation feasible, actively contribute to the work themselves, and form long-term relationships for future research.[25–29] Fundamentally, addressing representation involves showing communities, through words and actions, that their interests are inherently valuable and consistently addressed as part of a relationship that extends beyond the bounds of recruitment for an individual research study.

Group harms and implications for communities

Group harms are damages or injury, tangibly experienced or perceived, that impact the welfare interests of a group and its people.[12–14,30] There are many ways research studies might cause harm to groups. Genetic information can shed light on the ancestral origins, migration patterns, and demographic history of populations and thereby challenge cultural and cosmological beliefs and can sometimes have economic or legal impact on a group's claims or attributions. For example, genetic information was extracted from blood samples collected from members of the Havasupai Tribe living in Arizona for a diabetes study and was used in subsequent studies that concluded the group's ancestral roots extended to Asia.[31–33] Furthermore, there is evidence that researchers intended to use the samples for studies on schizophrenia, and research team members were told to withhold this information from the Tribe.[34] These conclusions and misuses of samples were harmful to the Tribe because the migration research conflicted with their own understanding of their origins and could be used in a legal context to contest the Havasupai Tribe's claim to their tribal lands. Both the migration research and schizophrenia research were seen as stigmatizing and exacerbating stereotypes about relatedness. The Tribe never received benefits from their participation nor were directly informed of the research results and its implications. The Tribe sued, and eventually the case ended in a settlement in which the Tribe received monetary compensation, and blood samples were returned.[31]

All too often, however, group harms are of a more subtle nature, and the wrongs created are ones that outsiders to the group would have trouble recognizing. For example, unintended harms can be caused by descriptions about the community that are not in alignment with the way the community views themselves. Members of the Deaf community, for example, highlight that being deaf represents one dimension of human diversity that need not be understood as a disease or disability, and that Deaf culture and language provide meaning and identity for its members in ways analogous to the culture provided within other minoritized groups.[35] Research publications that, for example, frame deafness as a loss of quality of life are insensitive and perpetuate harms to the Deaf community.

Members of minoritized communities will understandably be reluctant to participate in research that could create these types of harms. Addressing these concerns, however, requires bi-directional learning and communication. Fundamentally, communities need reassurance that the proposed research would not cause group harms, either in the immediate or long-term future. Researchers need to be forward thinking to minimize the chances that harms will come to the community as a result of such factors as long-term storage of samples, unrestricted secondary uses of data, and cycles of extractive research where few benefits return to the community.[36] In order to fulfill this responsibility, researchers need to demonstrate that they understand, and respect, the community's perspectives on potential group harms.

Meeting this expectation requires that researchers approach communities with humility, including cultural humility. With the understanding that even though researchers come to the community with ideas about research that they believe would be beneficial, they might need to accept that what they have in mind can instead bring about group harms.[30] Because of factors such as individualism, ableism, and systemic racism, researchers who are outsiders will often struggle to share the perspective of the community on the potential for group harms. Therefore, building trust between the community and researchers requires creating the space and time for listening. For researchers, this means listening to the community's perspective with enough humility to understand that the community itself is best positioned to anticipate group harms; for members of the community, it means learning about the researchers and what their resources might offer to the community. Researchers are likely to need to consider their own inherent biases, including the assumptions that they bring to the conversation that might make it difficult for them to hear what the community has to say. The inclusion of community members as members of the research team is one strategy that can be particularly effective for establishing the bi-directional learning and communication process that is needed to understand and avoid potential group harms.[25]

A common presumption in genetic and genomic research is that informed consent inherently encompasses these issues and that receipt of a signature assures community acceptance of the provisions included in it. Although consent is a necessary element of minimizing harms and maximizing voluntariness of research, consent alone is insufficient. The process of informed consent for research participation serves ethical, legal, and regulatory requirements, grounded in respect for persons and promotion of participant autonomy. However, individual members of a particular community might not be well-positioned to foresee group harms with the information conveyed during an informed-consent conversation. For some communities, traditional practices of informed consent are not enough and can be a source of misunderstanding and controversy because, in many cultural contexts, family- and community-centered values are emphasized, whereas individual autonomy is de-emphasized. Community engagement, and even community approvals (e.g., sign-off from community leaders or representatives), serve a complementary role to individual informed consent in some instances. When researchers engage certain communities, both are needed to help ensure that research with these groups avoids causing harms.

Approaches to community engagement

Given these considerations, it is clear that community engagement is an important way that the genetics and genomics research community can begin to address underrepresentation while avoiding the creation or exacerbation of group harms. As with many components of research, however, the "devil is in the details."

Taken as a whole, community engagement is "the process of working collaboratively with groups of people who are affiliated by geographic proximity, shared interests, or similar situations with respect to issues affecting their well-being."[2] Community engagement in research can take on many forms, many of which involve sharing information, developing tailored research questions, discussing concerns or expectations about research goals and processes, and fostering a dialogue between researchers and community members in an effort to conduct research with greater community impact and participation. The various approaches to community engagement contrast with *participant-engagement* approaches that aim to elicit the views about project progress, participant retention, and quality control of research practices from individuals *already recruited* into a study.[37]

Over the last few decades, one of the better-known community-engaged research approaches has been *community-based participatory research* (CBPR). This approach seeks to equitably involve all stakeholders, including researchers, community members, and community organizations, as partners in the development and implementation of a research program.[38–41] The CBPR model promotes collaborative partnerships throughout all aspects of the research process, including formulation of research questions and interpretation of study data. Many of the community-engagement

Table 1. Guidance for genetics and genomics researchers on adopting community-engaged research approaches throughout the research lifecycle

Preparatory to research: Making introductions	
Learn about the community	Learn about the community through local experiences, such as reading or listening to community news sources and attending local events.
Learn about the culture	Learn about cultural perspectives and beliefs on samples, DNA, relatedness, and other concepts; ensure research personnel have opportunities for learning about the community as well as building skills related to cultural humility.
Identify community partners and liaisons familiar to the community	There are numerous approaches that could work for identifying partners that are familiar to and trusted by community members. Institutional community relations offices, community organizations, and colleagues who have previously done community engagement can all be useful, among other approaches.
Work with community partners to address preliminary issues	Identify types of research that are consistent with the values and priorities of the community; explore appropriate use of language describing the community and how they would like to be referred to and better understand diversity internal to the community.
Preparatory to research: Building partnerships	
Identify concerns	Actively solicit and identify community concerns to be addressed. When they cannot be addressed, respect and defer to communities' views.
Co-develop plans	Develop plans in conjunction with the community about potential benefits of participating, expected tangible outcomes, plans for frequency of researcher engagement throughout the research, and plans to mitigate any potential harms.
Develop long-term goals and processes for engagement	Discuss engagement, relationship building, and future research with the local community as a long-term commitment before the beginning and after the end of any particular research goal/study/phase.
Discuss and develop metrics to evaluate the community-engagement effort	Evaluation efforts should take place throughout the lifecycle of engaged research. It is therefore important to plan the evaluation approach early in the process.
Form collaborations	Collaborate with people who have experience in engaging with the community.
Use resources	Utilize existing resources available online or in print, as well as university or institutional programs, that provide support for community-engagement activities.
Train community	Explore opportunities to train and empower community members to conduct parts of the research and to compensate them.
Develop resources	Access or develop community-appropriate resources about topics relevant to the research (e.g., text, images, or media), including resources in the language specific to the community.
Discuss data	Discuss ownership, privacy of results and data, and publications with research participants.
Plan for communication	Work with communities to inform how research findings will be communicated both to the community and to external audiences to maximize community benefit and impact and to avoid creating or exacerbating group harms.
Plan to return results	Anticipate return of results, including medically actionable results, to research participants and/or local healthcare providers in ways that are culturally appropriate and consider what is needed to properly equip participants or providers for a meaningful return-of-results process.
Share benefits	Explore opportunities to share the benefits of research, for example by returning results or helping the community identify and access resources to address gaps identified in the research.
During research	
Report findings	Report relevant research findings during the research study in a culturally appropriate and accessible manner to individual participants or the broader community.
Seek input	When appropriate, seek community input on ongoing processes or methodologic changes in data collection, analysis, interpretation, and reporting.
Engage community	Continue to engage the community throughout the research study, during the execution and optimization of the research plan.
Evaluate harms	Evaluate appropriateness of research from the perspective of potential unforeseen group harms not considered in the planning stage and that might arise during the research process.
Assess project	Assess strengths and weaknesses of the community-engagement process to remedy challenges and improve practice in the long term.

(*Continued on next page*)

Table 1. ***Continued***

After research	
Interpret results	Engage the community for appropriate interpretation (member checking) and dissemination of results in a manner that is consistent with the values and views of the community to provide benefit beyond scientific publications.
Consult community	Ask communities how they would like to be named, or referred to, in publications and other media, and about the potential for authorship or acknowledgement.
Return results	Enact an accessible and culturally sensitive plan for returning research results, including medically actionable results, to participants and/or healthcare providers.
Evaluate the community-engagement effort	Employ processes to elicit feedback on the success of the engagement approach.[59,60]
Reinforce long-term commitments	Engage the community in a long-term commitment with clear plans for benefit sharing before the beginning and after the end of any particular research goal, study, or phase.
Nurture relationships	Maintain long-term relationships with the community through longitudinal resources available to the community.

practices utilized in genetics and genomics research—outreach, consultation, collaboration, and shared leadership—are rooted in some of the same principles as CBPR but adapt the approach to meet the needs of the particular project. One principle from CBPR that is particularly important to community engagement in genetics and genomics research is the recognition of the collective expertise and wisdom that community members bring to shape research goals on the basis of their priorities and lived experiences. Another key principle of CBPR that is broadly applicable is that of addressing injustice. Community-engagement practices can help identify community perspectives on how best to address justice concerns, including ensuring access to the downstream benefits of research within underserved communities and assessing the possible distribution of gains or profits derived from the use of human genetic resources to those who contributed through their data, biosamples, or other types of effort. Benefit sharing of this type should focus on transparency, equitable distribution of benefits to research participants, and the provision of benefits to those who may lack reasonable access to the products and services that resulted from their contribution.[5,42,43]

Meaningful community relationships

Meaningfully engaging community perspectives and ensuring benefit sharing hinges on researchers' commitment to long-term relationships with these communities. Short-term relationships with underrepresented groups risk creating or perpetuating group harms and mistrust. Neglecting benefit sharing with the communities in which the research is performed can be considered extractive and exploitative, an approach to research that has been described as "helicopter" or "colonial.[44–47] These approaches are harmful to both communities and the goals of the broader scientific community because they can exacerbate mistrust of genomics research and decrease its scientific and social value. Maintaining long-term relationships can be especially challenging when researchers are not members of the community or when they reside far away. Researchers can address this challenge by engaging and establishing professional collaborations with community-based researchers or leaders from the originating communities as major stakeholders in collaborative research efforts. This approach can benefit the research by leveraging the experience and knowledge of the local community leaders or researchers about their communities while increasing the probability of long-term engagement with the communities through local members of the research group.

Limitations in time and resources are also important barriers to meaningful community engagement.[17,21,48] Some funding sources might not consider community engagement as part of the research plan and thus might not allocate funding and time for these activities. This lack of emphasis on community engagement can also be exacerbated by the practices of scientific journals, which might not be receptive to manuscripts that describe community-engagement activities and their role in the associated research. Encouraging funders and publishers to be cognizant of the importance of meaningful community-engagement efforts in genetics and genomics research, as well as the cost and effort required, will most likely increase the number of researchers and projects that engage in these activities.

Community members also face limitations on time and resources. Because of systemic inequities, community members often experience lack of childcare, inflexible work schedules, and logistical barriers such as lack of transportation. In addition, researchers might utilize inaccessible study locations or communication methods that fit poorly with the needs of the community. All of these factors can dissuade community members from participating in community-engagement activities to support research.[26,49] Just as importantly, some community members will have other priorities and commitments and might simply not be interested in the proposed research. To address some of these issues, engagement and study plans should assess barriers and potential forms of compensation with community members and ensure that the study design and implementation are respectful of their time and commitment. Creative ways to give back include helping community organizations with administrative tasks, supporting health promotion activities, and providing education resources.

Guidance for researchers

Genomics-research-related community-engagement efforts have been conducted in a variety of contexts and have employed a range of strategies. However, there is no one right way to conduct community engagement, and not all methodologies are transferable to every research project, population, and set up.[20,50–56] Nevertheless, certain aspects of community engagement are important to consider throughout the research cycle. Table 1 highlights the collaborative nature of research with the community of interest and outlines some of the steps that will facilitate respectful engagement and ultimately more successful research efforts. These recommendations are intended as a general guide and include considerations prior to conducting the research, during the research project, and after the conclusion of research processes. Other groups, such the Patient-Centered Outcomes Research Institute (PCORI), have provided detailed rubrics that might also be helpful to researchers seeking to develop an engagement strategy.[57] Ideally, there ought to be continuous communication with the research participant community throughout the research cycle, including after the study is completed.[51,58]

Conclusions

Ensuring that the benefits of participating in human genetics and genomics research are realized requires careful attention to minimize harms while actively seeking ways to maximize benefits. This guidance intends to advance community-engagement approaches by promoting bidirectional engagement between human genetics and genomics researchers and communities to pursue that vision successfully. Researchers in human genetics and genomics have the critical opportunity to utilize the methods of community-engaged research in their own work. This shift will help ASHG members build lasting community relationships that involve engaging in discourse about the research and its findings. It is with these types of relationships that researchers can create the opportunity to avoid group harms, engage in benefit sharing, and address health disparities, ultimately contributing to fulfilling the vision of ASHG that people everywhere will realize the benefits of human genetics and genomics research.

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References

1. Lavizzo-Mourey, R.J., Besser, R.E., and Williams, D.R. (2021). Understanding and Mitigating Health Inequities - Past, Current, and Future Directions. N. Engl. J. Med. *384*, 1681–1684. https://doi.org/10.1056/NEJMp2008628.
2. Clinical and Translational Science Awards Consortium; and Community Engagement Key Function Committee Task Force on the Principles of Community Engagement (2011). Principles of Community Engagement, Second Edition (Centers for Disease Control and Prevention). https://www.atsdr.cdc.gov/communityengagement/pdf/PCE_Report_508_FINAL.pdf.
3. Holzer, J.K., Ellis, L., and Merritt, M.W. (2014). Why we need community engagement in medical research. J. Investig. Med. *62*, 851–855. https://doi.org/10.1097/jim.0000000000000097.
4. Schroeder, D. (2009). Justice and Benefit Sharing Indigenous Peoples, Consent and Benefit Sharing (Springer).
5. Dauda, B., and Dierickx, K. (2013). Benefit sharing: an exploration on the contextual discourse of a changing concept. BMC Med. Ethics *14*, 36. https://doi.org/10.1186/1472-6939-14-36.
6. Royal, C.D., Novembre, J., Fullerton, S.M., Goldstein, D.B., Long, J.C., Bamshad, M.J., and Clark, A.G. (2010). Inferring genetic ancestry: opportunities, challenges, and implications. Am. J. Hum. Genet. *86*, 661–673. https://doi.org/10.1016/j.ajhg.2010.03.011.
7. Tervalon, M., and Murray-García, J. (1998). Cultural humility versus cultural competence: a critical distinction in defining physician training outcomes in multicultural education. J. Health Care Poor Underserved *9*, 117–125. https://doi.org/10.1353/hpu.2010.0233.
8. Greene-Moton, E., and Minkler, M. (2020). Cultural Competence or Cultural Humility? Moving Beyond the Debate. Health Promot. Pract. *21*, 142–145. https://doi.org/10.1177/1524839919884912.
9. Flanagin, A., Frey, T., Christiansen, S.L.; and AMA Manual of Style Committee (2021). Updated Guidance on the Reporting of Race and Ethnicity in Medical and Science Journals. JAMA *326*, 621–627. https://doi.org/10.1001/jama.2021.13304.
10. Stamper, K. (2019). Why We Confuse Race and Ethnicity: A Lexicographer's Perspective. Conscious Style Guide. https://consciousstyleguide.com/why-we-confuse-race-ethnicity-lexicographers-perspective/.
11. Borrell, L.N., Elhawary, J.R., Fuentes-Afflick, E., Witonsky, J., Bhakta, N., Wu, A.H.B., Bibbins-Domingo, K., Rodríguez-Santana, J.R., Lenoir, M.A., Gavin, J.R., 3rd., et al. (2021). Race and Genetic Ancestry in Medicine - A Time for Reckoning with Racism. N. Engl. J. Med. *384*, 474–480. https://doi.org/10.1056/NEJMms2029562.
12. McGregor, J.L. (2007). Population genomics and research ethics with socially identifiable groups. J. Law Med. Ethics *35*, 356–370. https://doi.org/10.1111/j.1748-720X.2007.00160.x.
13. Yu, J.H., and Juengst, E. (2020). Do Groups Have Moral Standing in Unregulated mHealth Research? J. Law Med. Ethics *48*, 122–128. https://doi.org/10.1177/1073110520917037.
14. Hausman, D.M. (2007). Group risks, risks to groups, and group engagement in genetics research. Kennedy Inst. Ethics J. *17*, 351–369. https://doi.org/10.1353/ken.2008.0009.
15. Ioannidis, J.P.A., Powe, N.R., and Yancy, C. (2021). Recalibrating the Use of Race in Medical Research. JAMA *325*, 623–624. https://doi.org/10.1001/jama.2021.0003.

16. National Research Council (2004). Measuring Racial Discrimination (The National Academies Press).
17. Lemke, A.A., and Harris-Wai, J.N. (2015). Stakeholder engagement in policy development: challenges and opportunities for human genomics. Genet. Med. *17*, 949–957. https://doi.org/10.1038/gim.2015.8.
18. Braveman, P.A., Arkin, E., Proctor, D., Kauh, T., and Holm, N. (2022). Systemic And Structural Racism: Definitions, Examples, Health Damages, And Approaches To Dismantling. Health Aff. *41*, 171–178. https://doi.org/10.1377/hlthaff.2021.01394.
19. Nooruddin, M., Scherr, C., Friedman, P., Subrahmanyam, R., Banagan, J., Moreno, D., Sathyanarayanan, M., Nutescu, E., Jeyaram, T., Harris, M., et al. (2020). Why African Americans say "No": A Study of Pharmacogenomic Research Participation. Ethn. Dis. *30*, 159–166. https://doi.org/10.18865/ed.30.S1.159.
20. Hiratsuka, V.Y., Hahn, M.J., Woodbury, R.B., Hull, S.C., Wilson, D.R., Bonham, V.L., Dillard, D.A., Alaska Native Genomics Research Workshop Group, Beckel-Mitchener, A.C., Blome, J., et al. (2020). Alaska Native genomic research: perspectives from Alaska Native leaders, federal staff, and biomedical researchers. Genet. Med. *22*, 1935–1943. https://doi.org/10.1038/s41436-020-0926-y.
21. Martinez, P., Cummings, C., Karriker-Jaffe, K.J., and Chartier, K.G. (2017). Learning from Latino voices: Focus Groups' Insights on Participation in Genetic Research. Am. J. Addict. *26*, 477–485. https://doi.org/10.1111/ajad.12531.
22. Bussey-Jones, J., Garrett, J., Henderson, G., Moloney, M., Blumenthal, C., and Corbie-Smith, G. (2010). The role of race and trust in tissue/blood donation for genetic research. Genet. Med. *12*, 116–121. https://doi.org/10.1097/GIM.0b013e3181cd6689.
23. Need, A.C., and Goldstein, D.B. (2009). Next generation disparities in human genomics: concerns and remedies. Trends Genet. *25*, 489–494. https://doi.org/10.1016/j.tig.2009.09.012.
24. Popejoy, A.B., and Fullerton, S.M. (2016). Genomics is failing on diversity. Nature *538*, 161–164. https://doi.org/10.1038/538161a.
25. Lemke, A.A., Choi, S.H., Dang, Q.T., Dang, V., and Yu, J.H. (2022). Assessing Vietnamese American patient perspectives on population genetic testing in primary care: a community engaged approach. HGG Advances. https://doi.org/10.1016/j.xhgg.2022.100134.
26. Sabatello, M., Chen, Y., Zhang, Y., and Appelbaum, P.S. (2019). Disability inclusion in precision medicine research: a first national survey. Genet. Med. *21*, 2319–2327. https://doi.org/10.1038/s41436-019-0486-1.
27. Lewis, K.L., Turbitt, E., Chan, P.A., Epps, S., Biesecker, B.B., Erby, L.A.H., Fasaye, G.A., and Biesecker, L.G. (2021). Engagement and return of results preferences among a primarily African American genomic sequencing research cohort. Am. J. Hum. Genet. *108*, 894–902. https://doi.org/10.1016/j.ajhg.2021.04.002.
28. Claw, K.G., Anderson, M.Z., Begay, R.L., Tsosie, K.S., Fox, K., Garrison, N.A.; and Summer internship for INdigenous peoples in Genomics SING Consortium (2018). A framework for enhancing ethical genomic research with Indigenous communities. Nat. Commun. *9*, 2957. https://doi.org/10.1038/s41467-018-05188-3.
29. Kobayashi, Y., Boudreault, P., Hill, K., Sinsheimer, J.S., and Palmer, C.G.S. (2013). Using a social marketing framework to evaluate recruitment of a prospective study of genetic counseling and testing for the deaf community. BMC Med. Res. Methodol. *13*, 145. https://doi.org/10.1186/1471-2288-13-145.
30. Miller, P.S., and Levine, R.L. (2013). Avoiding genetic genocide: understanding good intentions and eugenics in the complex dialogue between the medical and disability communities. Genet. Med. *15*, 95–102. https://doi.org/10.1038/gim.2012.102.
31. Harmon, A. (2010). Indian Tribe Wins Fight to Limit Research of Its DNA. New York Times.
32. Mello, M.M., and Wolf, L.E. (2010). The Havasupai Indian trive case-lessons for research involving stored biologic samples. N. Engl. J. Med. *363*, 204–207.
33. Garrison, N.A. (2013). Genomic Justice for Native Americans: Impact of the Havasupai Case on Genetic Research. Sci. Technol. Human Values *38*, 201–223. https://doi.org/10.1177/0162243912470009.
34. Hart, S., and Sobraske, K. (2003). Investigative Report Concerning the Medical Genetics Project at Havasupai (Reserves: Photocopy of investigative report available at the Arizona State University Ross-Blakey Law Library).
35. Garofalo, D.C., Rosenblum, H.A., Zhang, Y., Chen, Y., Appelbaum, P.S., and Sabatello, M. (2022). Increasing inclusivity in precision medicine research: Views of deaf and hard of hearing individuals. Genet. Med. *24*, 712–721. https://doi.org/10.1016/j.gim.2021.11.015.
36. Hudson, M., Garrison, N.A., Sterling, R., Caron, N.R., Fox, K., Yracheta, J., Anderson, J., Wilcox, P., Arbour, L., Brown, A., et al. (2020). Rights, interests and expectations: Indigenous perspectives on unrestricted access to genomic data. Nat. Rev. Genet. *21*, 377–384. https://doi.org/10.1038/s41576-020-0228-x.
37. Childerhose, J.E., Finnila, C.R., Yu, J.H., Koenig, B.A., McEwen, J., Berg, S.L., Wilfond, B.S., Appelbaum, P.S., and Brothers, K.B. (2019). Participant Engagement in Translational Genomics Research: Respect for Persons-and Then Some. Ethics Hum. Res. *41*, 2–15. https://doi.org/10.1002/eahr.500029.
38. Israel, B.A., Schulz, A.J., Parker, E.A., and Becker, A.B. (1998). Review of community-based research: assessing partnership approaches to improve public health. Annu. Rev. Public Health *19*, 173–202. https://doi.org/10.1146/annurev.publhealth.19.1.173.
39. Wallerstein, N.D., Bonnie, Oetzel, J., and Minkler, M. (2017). Community-Based Participatory Research for Health: Advancing Social and Health Equity (Jossey-Bass).
40. Wallerstein, N., and Duran, B. (2010). Community-based participatory research contributions to intervention research: the intersection of science and practice to improve health equity. Am. J. Public Health *100* (*Suppl 1*), S40–S46. https://doi.org/10.2105/ajph.2009.184036.
41. Israel, B.A., Coombe, C.M., Cheezum, R.R., Schulz, A.J., McGranaghan, R.J., Lichtenstein, R., Reyes, A.G., Clement, J., and Burris, A. (2010). Community-based participatory research: a capacity-building approach for policy advocacy aimed at eliminating health disparities. Am. J. Public Health *100*, 2094–2102. https://doi.org/10.2105/ajph.2009.170506.
42. Schroeder, D. (2007). Benefit sharing: it's time for a definition. J. Med. Ethics *33*, 205–209. https://doi.org/10.1136/jme.2006.016790.
43. Laird, S., Wynberg, R., Rourke, M., Humphries, F., Muller, M.R., and Lawson, C. (2020). Rethink the expansion of access

and benefit sharing. Science *367*, 1200–1202. https://doi.org/10.1126/science.aba9609.

44. Atutornu, J., Milne, R., Costa, A., Patch, C., and Middleton, A. (2022). Towards Equitable and Trustworthy Genomics Research. EBioMedicine *76*. https://doi.org/10.1016/j.ebiom.2022.103879.
45. Reardon, J., and TallBear, K. (2012). Your DNA is our history": Genomics, anthropology, and the construction of whiteness as property. Curr. Anthropol. *53*, S233–S245.
46. Rose, J. (2016). Biopiracy: When Indigenous Knowledge is Patented for Profit. The Conversation. https://theconversation.com/biopiracy-when-indigenous-knowledge-is-patented-for-profit-55589.
47. Breske, A. (2018). Biocolonialism: Examining Biopiracy, Inequality, and Power. Spectra *6*, 58–73. https://doi.org/10.21061/spectra.v6i2.a.6.
48. Lemke, A.A., Wu, J.T., Waudby, C., Pulley, J., Somkin, C.P., and Trinidad, S.B. (2010). Community engagement in biobanking: Experiences from the eMERGE Network. Genom. Soc. Policy *6*, 50. https://doi.org/10.1186/1746-5354-6-3-50.
49. Sabatello, M., Blake, L.A., Chao, A., Silverman, A., Ovadia Mazzoni, R., Zhang, Y., Chen, Y., and Appelbaum, P.S. (2019). Including the blind community in precision medicine research: findings from a national survey and recommendations. Genet. Med. *21*, 2631–2638. https://doi.org/10.1038/s41436-019-0533-y.
50. Lemke, A.A., Halverson, C., and Ross, L.F. (2012). Biobank participation and returning research results: perspectives from a deliberative engagement in South Side Chicago. Am. J. Med. Genet. *158a*, 1029–1037. https://doi.org/10.1002/ajmg.a.34414.
51. Agongo, G., Debpuur, C., Amenga-Etego, L., Nonterah, E.A., Kaburise, M.B., Oduro, A., Ramsay, M., and Tindana, P. (2021). Community engagement and feedback of results in the H3Africa AWI-Gen project: Experiences from the Navrongo Demographic and Health Surveillance site in Northern Ghana. AAS Open Res. *4*, 15. https://doi.org/10.12688/aasopenres.13081.1.
52. Association of American Medical Colleges Center for Health Justice (2022). Community Engagement, Precision Medicine, and Native Health. https://www.aamchealthjustice.org/resources/community-engagement-toolkits/community-engagement-precision-medicine-native-health.
53. Kaladharan, S., Vidgen, M.E., Pearson, J.V., Donoghue, V.K., Whiteman, D.C., Waddell, N., Pratt, G.; and GenetiQs Study (2021). Ask the people: developing guidelines for genomic research with Aboriginal and Torres Strait Islander peoples. BMJ Glob. Health *6*, e007259. https://doi.org/10.1136/bmjgh-2021-007259.
54. Rotimi, C., Leppert, M., Matsuda, I., Zeng, C., Zhang, H., Adebamowo, C., Ajayi, I., Aniagwu, T., Dixon, M., Fukushima, Y., et al. (2007). Community engagement and informed consent in the International HapMap project. Community Genet. *10*, 186–198. https://doi.org/10.1159/000101761.
55. Faure, M.C., Wonkam, A., and De Vries, J. (2020). Using the Drama of DNA Approach to Community Engagement in Genomic Research in South Africa: Experiences and Lessons Learnt. AAS Open Res. *3*, 1. https://doi.org/10.12688/aasopenres.13045.1.
56. Beaton, A., Hudson, M., Milne, M., Port, R.V., Russell, K., Smith, B., Toki, V., Uerata, L., Wilcox, P., Bartholomew, K., and Wihongi, H. (2017). Engaging Māori in biobanking and genomic research: a model for biobanks to guide culturally informed governance, operational, and community engagement activities. Genet. Med. *19*, 345–351. https://doi.org/10.1038/gim.2016.111.
57. PCORI (Patient-Centered Outcomes Research Institute) (2016). Engagement Rubric for Applicants. http://www.pcori.org/sites/default/files/Engagement-Rubric.pdf.
58. Trinidad, S.B., Ludman, E.J., Hopkins, S., James, R.D., Hoeft, T.J., Kinegak, A., Lupie, H., Kinegak, R., Boyer, B.B., and Burke, W. (2015). Community dissemination and genetic research: moving beyond results reporting. Am. J. Med. Genet. *167*, 1542–1550. https://doi.org/10.1002/ajmg.a.37028.
59. Bowen, D.J., Ackermann, N., Thompson, V.S., Nederveld, A., and Goodman, M. (2022). A Study Examining the Usefulness of a New Measure of Research Engagement. J. Gen. Intern. Med. *37*, 50–56. https://doi.org/10.1007/s11606-021-06993-1.
60. Clinical and Translational Science Awards Consortium; and Community Engagement Key Function Committee Task Force on the Principles of Community Engagement (2011). Chapter 7: Program Evaluation and Evaluating Community Engagement. In Principles of Community Engagement, Second Edition (Atlanta, Centers for Disease Control and Prevention), pp. 163–178.