Race, Ancestry, and Representation: The Role of Genetic Medicine

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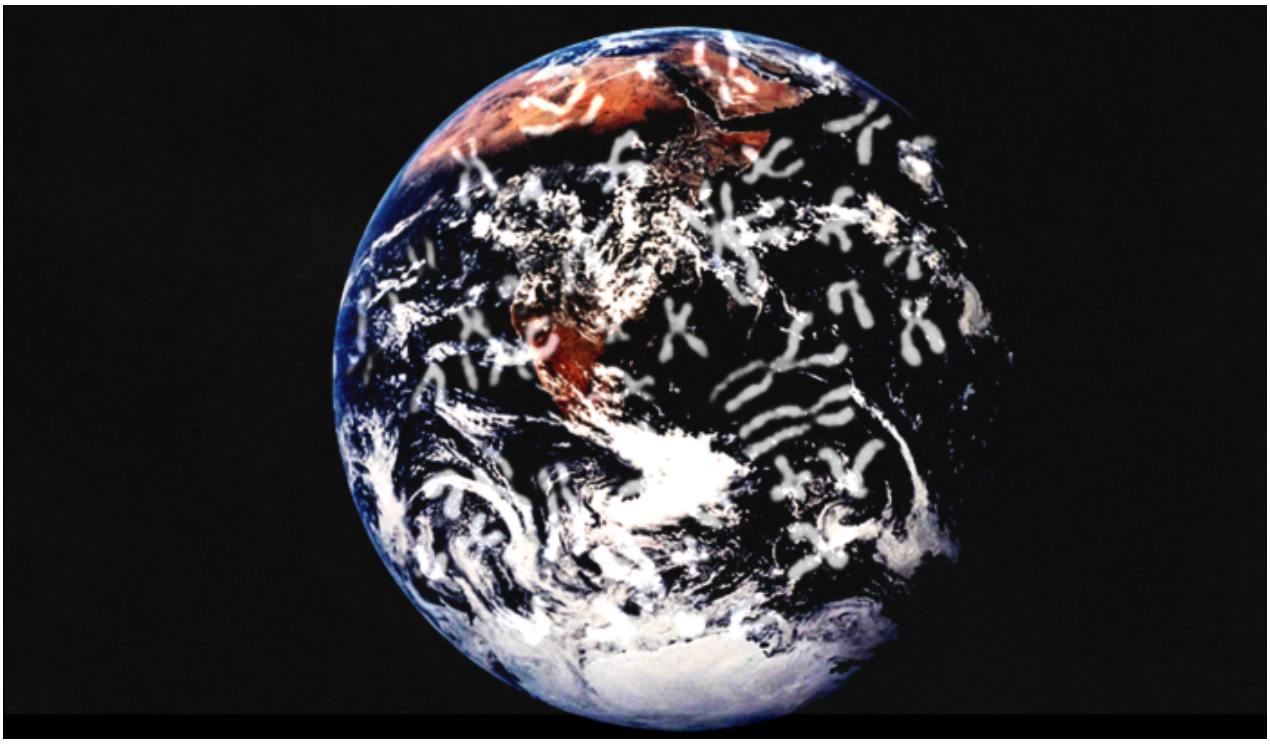
Race is a social construct, but representation matters in genetic studies — and on that front, we have a long way to go. By Tala Berro



By Tala Berro

I am a Palestinian American. On the census, under the racial category, I am instructed to mark the box labeled “white.” Why am I classified as white? On doing a deeper dive about this, I found out that the older Arab generations wanted this classification in order to better assimilate. This did help many lighter-skinned Arabs to assimilate, yet there is still a history of people from SWANA (Southwest Asia, North Africa) and Muslim communities being heavily surveilled. The politics surrounding the categorization of my “race” brings up for me the reminder that race is a social construct, not a product of biology.

The pernicious misinformation around race being biological has persisted for far too long. Half of medical students surveyed in 2016 endorsed at least one false belief that race leads to biological differences, such as that Black people’s nerve endings are less sensitive, white people have larger brains, or Black couples are more fertile. Similar myths are coming out this year in relation to how COVID-19 is disproportionately affecting BIPOC (Black, indigenous, and people of color) communities.

Being a social construct is not the same as being imaginary. In the United States, we see an array of disparities in health outcomes along racial lines. As Dr. Camara Jones notes: “Race doesn’t put you at higher risk. *Racism* puts you at higher risk. It does so

through two mechanisms: People of color are more infected because we are more exposed and less protected. Then, once infected, we are more likely to die because we carry a greater burden of chronic diseases from living in disinvested communities with poor food options [and] poisoned air and because we have less access to health care."

To be honest, I didn't grapple with this concept until I heard Dr. Tina Sacks speak on this subject at the National Society of Genetic Counseling (NSGC) Annual Education Conference. Dr. Sacks is the author of *Invisible Visits*, which highlights how discrimination continues to affect Black women of all socioeconomic levels in healthcare settings. While race is a construct unsubstantiated by biological data as the cause of disparities, there are real social consequences associated with race.

There are also real genetic variants between people of different ancestries, but it is based on differences among local populations and regions, not race. As Vivian Chou explains: "Ultimately, there is so much ambiguity between the races, and so much variation within them, that two people of European descent may be more genetically similar to an Asian person than each other." In fact, there is greater genetic diversity among the ~2000 African ethno-linguistic groups than with the rest of the global population. Overall, the genomics field stands to gain immense knowledge from studying African populations, and several initiatives support

African investigators conducting this important work.

We also see racial disparities in genetic research. Nearly three-quarters of all genome-wide association studies done between 2005 and 2018 came from people living in just three countries: the United Kingdom, the United States, and Iceland. To make new genetic discoveries and have true knowledge of genetic variation, we need to include people of all backgrounds in genetic research, in all levels — PIs, researchers, trainees and participants. I recall a NSGC workshop about variants of uncertain significance (VUS) where an audience member asked about VUS results in patients who do not have European ancestry. The speaker noted that this was to be expected, and that there was nothing to do about it. This response by our field is unjust and unacceptable.

As a Palestinian woman, I think about the lack of genetic information from individuals with SWANA ancestry. But even more importantly, I think about how this lack of diversity may further exacerbate the medical disparities we already see due to unconscious bias, racism and other forms of discrimination. We see this happening when we dismiss someone with a genetic condition because they are not the “expected race” to have that disorder. This expectation is rooted in our limited understanding of genetic variation, which is predominately determined and biased by the preponderance of data from people of European ancestry and lack of data specifically from individuals of African

ancestry.

Increasing diversity in genetic studies is one pertinent first step, and we must be mindful of how we go about addressing it. Black, indigenous, and other minority communities have good reason to be wary of medical researchers, and the medical field in general, between ongoing present-day health disparities — Black women in the U.S. are over three times more likely to die in childbirth than white women, regardless of income level — and our country's history of medical apartheid. Medical scholar Harriet Washington describes how racist pseudoscience remained mainstream in the American medical establishment well into the 20th century, built on false assumptions "that Black people were very, very different from whites, medically and biologically." From Henrietta Lacks to the Tuskegee Experiment to COVID-19 trials lacking minority representation when minorities are the ones most impacted by COVID-19, medical researchers have and continue to perpetuate harm upon Black Americans.

First Steps to Mitigate the Problem: The PeopleSeq Consortium

I began my role as genetic counselor and project manager at Mass General Brigham just two years ago. I was very eager to learn and grow as a professional, but also to see how I could bring my experiences as a minority into my position.

One of my roles within the Genomes2People (G2P) research group is project manager for the PeopleSeq Consortium. It is an NIH-funded study seeking to survey healthy adults who plan to, or have already received, their own genomic sequence information, and we currently partner with 15 various commercial and research avenues. Our hope is to be able to understand the experience of receiving this information as an ostensibly healthy person.

In the pilot phase of this project, less than one percent of participants self-identified as African American or Black. I was the first person of color on the operational team. This is not unusual for a genetics research team, especially one with genetic counselors.

As a first attempt to mitigate this disparity, we asked colleagues Dr. Tshaka Cunningham and genetic counselor Dr. Altovise Ewing to join our PeopleSeq team. We also applied for and received an NIH supplement to subsidize sequencing specifically for participants of African ancestry. To bring this subsidy to reality, Dr. Cunningham and Dr. Ewing spearheaded an event designed to provide in-depth information to community members about personal genome sequencing, insurance, privacy and what to expect from undergoing clinical sequencing and participating in the PeopleSeq survey study. At the end of the event, the audience members who were interested in pursuing their own genetic

sequencing were given the option to do so. This would involve pre- and post-test counseling and physician order as well, entirely separate from our PeopleSeq team.

While this one step in our one study is a micro-attempt at bringing in more Black collaborators and participants, it is an important first step for many studies to take. We are also undergoing similar steps in two of our newest projects. In PopSeq (Return of Genomic Results and Estimating Penetrance in Population-Based Cohorts), we are returning results to individuals who have undergone sequencing as part of the Jackson Heart Study, which is one of the largest studies of cardiovascular disease among African Americans. And in BabySeq2 (Implementation of Whole Genome Sequencing as Screening in a Diverse Cohort of Health Infants), we are working with an advisory board to recruit majority non-white participants from community-based clinics. Improving representation is necessary in order for the field of genetics to address its own role in medical apartheid.

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Genomes2People (G2P) is a program of Brigham and Women's Hospital, the Broad Institute and Harvard Medical School. Visit genomes2people.org for more and follow us on Twitter @Genomes2People.

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