Making Genomics as Diverse as Humanity

The lack of diversity in genomic research may be a call to fundamentally change the research enterprise.

By Robert C. Green

Medical research has a diversity problem. It happens through a combination of factors, from researchers' own uneven recruitment practices to many communities' mistrust of medical institutions, built both on historical atrocities and on present-day gaps in quality of medical care. A recent review of clinical trials around the globe found that 76% of participants were white and only 7% were Black. In the United States, the National Institutes of Health released a new strategic plan in response to persistent health disparities and those same communities' lack of representation in research.

In our own clinical trials, the Genomes2People Research Program has been more successful than most in achieving diversity among our participants; for example, the REVEAL Study on Alzheimer's Disease risks consistently included at least 20% individuals with African ancestry.

With clinical genomics, though, we face a different set of headwinds. What's different about genetics? Why do we find special challenges, not just for recruitment, but for the entire integration of genomics into medical science?

I had a conversation earlier this year with Dr. Evelynn Hammonds, Chair of the Department of the History of Science and Professor of History of Science and of African and African-American Studies at Harvard University. Dr Hammonds points out that genetics is sometimes discussed today in ways that reinscribe old ideas of genetic determinism, "the notion that there's something fundamentally biologically different between white groups of people and Black, brown, and Asian groups of people." We see this at times with DNA ancestry testing, sometimes misused as a way of putting people into racial and ethnic categories. It brings up echoes of the "one drop" rule of Jim Crow, in which any trace of African ancestry legally classified a person as "Negro."

"It's brought back a whole story that, many times in the history of medicine, we thought had been resolved."

As Dr. Hammonds points out, historical mistrust is also kept alive by ongoing disparities in medical care. "Many Black and brown people walk into a medical setting with the sense that it's just not going to turn out well. That they're not going to be treated with the kind of care that will allow them to actually be helped by these new technologies, genetics and genomics.

"There's a sense of skepticism: You've used your new technologies on us before and it hasn't turned out well. Why should we believe you now?"

Some researchers (and practitioners) get frustrated with this; but if we are actually listening, it sounds like a call to fundamentally change the research enterprise. To take seriously the same mindset shift that led us to say "research participants" instead of "research subjects."

I believe that an important step in this direction, especially for genomic research, is to normalize returning potentially life-saving research results to participants.

To many, this sounds obvious. Medical research can already feel akin to medical care, from a participant's perspective, and we've heard plenty of feedback along the lines of: *If you find something that can save my life, I expect you to tell me about it.* We've been returning results for years in some of our own trials, including over 2,600 people through a partnership with the Jackson Heart Study. Yet returning results remains a contentious practice, especially in genomic research.

"The research *should* be given back to participants, as part of the ethics of care," Dr. Hammonds agrees. "Particularly for populations with a long history of being used to produce new medical knowledge, but not having that knowledge actually serve them."

Other steps are needed, too. Researchers are often resistant, even resentful, toward requirements to have a certain percentage of people of color in their research recruitment pool; at the same time, they may view the participants themselves as "resistant" to participating. Here, Dr. Hammonds makes an important distinction: Sometimes, "resistance exists because there are real, absolute barriers to participation." She endorses the idea of paying participants to join a study, accounting for factors like transportation costs and taking time off work.

All of this overlaps with another fundamental issue, one we have also discussed in this space: "We have not solved the problem of why it's so difficult to increase the diversity of the population of medical *researchers*." The same can be said of related fields, including genetic counseling.

Returning research results, however controversial, may sound like a small piece of the puzzle. It could also be the next step forward in real, impactful efforts to diversify genomic research.

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