



# Genetic testing and results disclosure in diverse populations: what does it take?

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In their important paper, Shaibi and colleagues describe the challenges of returning actionable gene panel results to 500 low-income Latinx adults in a Federally Qualified Health Center (FQHC).<sup>1</sup> Using principles of community-based participatory research (CBPR),<sup>2</sup> the study team collaborated with community partners to create a culturally sensitive outreach strategy for returning results. Of the 500 individuals tested, nearly all had negative results returned by mail. Only 10 individuals had pathogenic or likely pathogenic results. Most received their results during in-person meetings with a genetic counselor (GC). The fastest time from consent to results disclosure for these ten participants was 507 days. Some could not be reached; one was too upset to receive results.

Why was it so difficult to return results in a timely manner? The authors describe missed appointments, low health literacy, lack of insurance, inadequate family history, and emotional distress. Answering this question writ large requires recognizing and addressing (not just measuring) social determinants leading to unjust racial, ethnic, and socioeconomic health disparities beyond this study. It also requires attending to research issues such as the need for pilot testing, community engagement, relationship building, research team diversity, flexibility, and addressing implicit bias. Based on literature review and research experience, the following strategies merit further consideration and research.

## PILOT TESTING

Pilot testing can determine feasibility and allow for early identification and modifications of problem areas.<sup>3</sup> In this case, it may have helped uncover, investigate, and alter protocols to address the unusually low P/LP rates, and challenges returning results before full implementation. During study development and piloting, study teams can also plan for situations researchers, clinicians, and community partners identify as potentially problematic at the outset, such as handling participants who may become despondent during return of results visits, participants with disabilities, limited English proficiency or health literacy, and producing

educational materials and resources participants can utilize after studies end. The piloting phase will also allow the team to complete all research development, so studies do not launch until critical procedures are tested and deemed ready to launch.

## CONTINGENCY PLANNING

To ensure successful, efficient results disclosure and participant follow up, it is important to develop contingency plans.<sup>4</sup> If piloted, deployed, and listed in study protocols, these plans provide teams a clear path for addressing deviations such as inability to complete results disclosure within a predetermined timeframe. Early identification of protocol deviations can trigger problem-solving meetings. Putting end dates on recontact can motivate teams and emphasize to all involved the urgency of returning results.

## BUILDING RELATIONSHIPS WITH STUDY PARTICIPANTS

Recruitment and retention are “a good invitation to a great party.”<sup>5</sup> It is essential to build relationships with participants from the outset, so they understand the importance of not only the research, but also of their contribution. Pressure to recruit participants rapidly could lead to inadequate attention building relationships and poor retention. Attending to relationships will allow research coordinators and GCs to learn barriers participants may have for follow up before problems arise (i.e., unreliable phones, housing instability). Studies should have detailed procedures to collect and update contact and alternative contact information, giving participants easy ways to reach study staff, emphasizing staff continuity, and identifying expanded options for recontact (e.g., people will be more likely to provide their cell number and open texts from people they trust). Documenting participants’ concerns and levels of engagement may guide future contacts.<sup>5</sup> A diverse and culturally competent study team, particularly one that reflects the demographics of the target population, has wide-ranging positive effects on participant engagement and follow up.<sup>6</sup>

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## RESEARCH TEAM FLEXIBILITY AND EXPANDING COMMUNICATION OPTIONS ARE IMPORTANT

Low resource communities have less reliable transportation, childcare, and work schedules with less flexibility and therefore narrower windows for interactions outside the weekday schedules study teams prefer. By asking participants to accommodate to our schedules, we send a message that we do not value their time and effort. Perhaps set up teams in which staff, GCs, and study space are available evenings and weekends. Teams should conduct virtual visits whenever possible, considering in-person visits only when absolutely necessary. In addition to phone calls or mailed letters, staying in contact with participants may be more effective through secure emails, text messages, reaching alternate contacts, sending certified letters, or intercepting participants at scheduled medical visits. At enrollment, staff should garner multiple ways to communicate, and preferred days and times for contact.

## REFRAMING SOCIAL DETERMINANTS OF HEALTH

Social determinants are sometimes considered barriers to research participation, or variables to measure and report on or control for in analyses; however, researchers should reframe them as a responsibility of the research team to identify and address. This may challenge, even obligate, researchers to alter how they conduct research. Should we offer genetic testing to participants who do not have insurance or other means to get follow-up testing or specialty care based on test results? Are challenges with language and health literacy participant problems, or societal problems requiring us to provide materials that meet their needs? If participants cannot afford transportation to get their research results, should we ask for this information in advance and either build this into the cost of testing, provide results through alternative means, or not offer testing that participants cannot get results for?

## STEREOTYPE CAUTION

There is a fine line between understanding and stereotyping a population. Communities underrepresented in research (i.e., non-White, low-income, non-English speaking) may be stereotyped as low literate, fatalistic, and viewing preventive care as a luxury.<sup>7</sup> We have not found this to be the case.<sup>5</sup> The sites where they receive care may be stereotyped as mere “safety nets.” However, FQHCs demonstrate equal or better performance on many quality measures, despite serving patients with more chronic diseases and greater socioeconomic complexity. Over 90% have interpreters available.<sup>8</sup> Challenging stereotypes can help study teams better understand and build relationships with potential participants, leaders of their communities, and the clinicians who care for them.

## GENETIC WORKFORCE TRAINING AND IMPLEMENTATION

There may be unintended process differences and racial bias in genetic counseling.<sup>9</sup> It may be beneficial to have

researchers, GCs, and staff who are from the communities they target, but diverse GCs are in limited supply, and lower-resource settings like FQHCs may not have any on staff. To provide diversity and access, GCs may not be necessary in all genetic encounters, and encounters can often be virtual. Studies comparing in-person to telephonic counseling, consenting, and results disclosure for actionable variants show noninferiority of telephonic discussions.<sup>10</sup> Studies employing trained laypersons from local communities carefully supervised by GCs show great promise, especially if focused in areas such as chronic disease risk and pharmacogenomics.<sup>11</sup>

## COMMUNITY-BASED PARTICIPATORY RESEARCH IS AN APPROACH, NOT A METHOD

As the authors noted, community stakeholders possess invaluable expertise regarding all aspects of the research process. These individuals should be considered partners, not passive advisors. Engaging such experts in developing the initial protocol, piloting, building relationships, meeting needs of participants, addressing challenges that inevitably arise, providing feedback if researchers misstep, and suggesting analyses may prove invaluable.<sup>2</sup> Our academic community team recruited (with 7% refusal) and retained (with 7% attrition) over 2000 African ancestry participants into a genetic testing trial in FQHCs and other practices in New York City in just two years. Lay staff overseen by GCs returned results in person, by phone or mail, with over 95% satisfaction, and while all participants were offered free in-person or telephone GC appointments, zero chose to have one.<sup>5</sup> In regular meetings with stakeholders (who we paid for their time), we shared data and problems, which they helped solve, every single time.

## RESEARCH EQUITY IS INTENTIONAL

Racism, classism, and historic research mishandling have created fragile relationships with low resource and nonwhite communities. Instead of lamenting that potential participants misunderstand, mistrust, and undervalue research, we can recognize and respect the legitimate challenges each individual faces taking part in research, their “earned skepticism” of research, and that their time, histories, ideas, and data are precious resources.<sup>2</sup> Researchers should challenge ourselves to consider that overcoming these challenges is our solemn responsibility. If we believe in the power of research to benefit human health, we must create positive research experiences for communities. By creating good invitations to great parties, showing diverse participants they are our priority not an afterthought, providing them with the results of the research (not just their individual results), we can avoid the “drive-by” or “helicopter” research people have experienced, in which they take part but derive no recognizable benefit. If we focus on these, we can build a community of diverse research partners and participants for the future.

In conclusion, many obstacles to research participation may be addressed through participatory planning, piloting,

flexibility, and an approach that acknowledges social and instrumental challenges. And, through carefully planned and explicitly studied implementation of genomic interventions, researchers can identify, preempt, and evaluate barriers to implementation of genomic medicine in diverse populations.

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### DISCLOSURE

C.R.H. declares no conflicts of interest.

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