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Should Researchers Share Genomic Results With Participants' Relatives?



By Robert C. Green, MD, MPH



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Your brother recently died of a rare form of cancer. You know that he had been participating in a study on the genetics of this cancer, and that as part of the study, researchers promised that they would send him his individual results. Those results would include his genetic risks related to this form of cancer, and it could also tell you, as his sibling, about your own risk. So you find a phone number for the study and call them up to ask for your brother's results.

Now let's say you're the researcher on the other end of this call. On the one hand, sharing this information might save the sibling's life. On the other hand, doesn't this open a whole can of worms? Researchers are required to protect the privacy of research participants. For starters, how do you know this caller is really the participant's sibling? That's only the first of several logistical issues — and all of this takes time and resources away from your research, especially if these requests become a regular occurrence.

When researchers discover something about a participant that could significantly affect that person's relatives, do the researchers have a duty to tell the relatives? Are the researchers bound to protect the participant's privacy even if the participant is deceased?

A group of researchers, clinicians, and ethicists from across the country had been struggling with these questions, so we formed a group, led by Susan Wolf, Barbara Koenig and Gloria Petersen, to tackle them and come up with some recommendations for researchers. We'll get to those recommendations in a minute, but first a little more explanation is in order.

In many types of human research, individual findings might not matter much to anyone besides the participants themselves. Genomic research is a bit different, though; first-degree biological relatives share 50% of the participant's genetic material. If researchers discover that a participant carries a gene linked to a dangerous but possibly preventable disease, they know that the participant's relatives may also carry the gene. Warning these relatives could save lives — yet this rarely happens. Why?

To begin with, study design and other logistics might make this very difficult. The samples may have been made anonymous in order to protect the participants' privacy, especially with something as sensitive as genomic information. Even if researchers have the participants' personal details, that contact information may be out of date; even if their contact info is up to date, tracking down each participant may be quite time-consuming and expensive—never mind trying to locate their relatives. Even if researchers can overcome the logistical hurdles, our recommendations (spoiler alert) discourage this sort of “active disclosure policy,” in which researchers initiate contact with relatives.

So, in the great majority of cases, cold-calling relatives isn't on the table. That leaves two main ways that a participant's genomic research results may reach their relatives.

First, if participants receive their results from a study (having agreed to this from the outset),

they are free to share these results with their relatives. Researchers might not be involved in this at all, or maybe they'll provide the participants with some guidance about how to explain their results to others. In our recommendations, we advised that researchers generally should not actively contact relatives, but should share information with the participants themselves, then urge participants to share their results with relatives. Generally speaking, the researcher's role—if they are even returning results to participants at all—is to disclose the results and let the participant take it from there.

Second—and this is where things get stickier—is what to do when a participant is debilitated or deceased, and when a participant's relatives contact the researchers. If the study was designed to include returning results, researchers may be able to share those results with participants' relatives as well. But researchers have a responsibility—legally and ethically—to protect their participants' privacy and confidentiality. Their duty is to the participant, not to the participant's relatives. This was a central point for us, both for the sake of research participants (who should be able to trust that their privacy will be respected) and researchers (whose primary job is to create knowledge, not to dispense medical advice).

Our conclusion: Researchers are not obligated to return a participant's results to relatives.

To be clear, though, we aren't saying that researchers shouldn't be involved in returning results to relatives — only that they aren't ethically obligated to do so. For researchers who do want to facilitate returning results to relatives who request them, we say (to paraphrase): Go ahead, but only if you are very sure you aren't doing anything the participants haven't agreed to.

That's what our recommendations largely boil down to:

1. Find out what the participant's preferences are.
2. Follow them.

Of course, it's slightly more complicated than that. This means getting participants' preferences if there is any possibility of returning results to relatives. It also means specifically asking about the participant's preferences for how results could be disclosed in the event of their death, and ideally having them identify a representative who could make decisions on their behalf if and when the participant isn't able.

We all agreed that participants (or their representatives) should control access to their

individual results. We also generally agreed that researchers should have a passive disclosure policy when it comes to sharing results with relatives: either let the participant share the information themselves, or if the researcher shares directly with the participant's relatives—having received clear approval from participants to do so—it should only be because those relatives requested it.

That said, we discussed a rare exception in which a researcher might be justified in actively reaching out directly to a participant's relatives. What if the researcher has found some genetic variant that is very likely to cause disease and also actionable — in other words, where telling relatives about this variant is likely to save someone from imminent harm? In this situation, would the researcher be ethically justified if they actively attempted to warn relatives?

Our group was split on this question. A majority felt that yes, in this rare instance, researchers could take the initiative and reach out to relatives. But some of the group held firm to the rule that only participants or their representatives should be able to control access to the participant's genomic results.

Researchers aren't always doctors — or if they are, providing patient care is not their role in this situation. Providing medical information to participants' relatives adds new logistical wrinkles, requires extra resources, and simply won't be feasible for many studies. But if a participant's relatives want the information, the researchers have a plan for how to share it, and the participant has consented, researchers may be ethically justified in sharing genomic research results with a participant's relatives.

After all, it could save lives.

Follow Robert C. Green, MD, MPH on Twitter: www.twitter.com/RobertCGreen



Robert C. Green, MD, MPH 

Medical Geneticist, Brigham and Women's Hospital and Harvard Medical School