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Returning genetic research results: study type matters

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Abstract

Aim—The return of individual genetic research results has been identified as one of the most pressing ethical challenges warranting immediate policy attention. We explored the practices and perspectives of genome-wide association studies (GWAS) investigators on this topic.

Materials & methods—Corresponding authors of published GWAS were invited to participate in a semistructured interview. Interviews (n = 35) were transcribed and analyzed using conventional content analysis.

Results—Most investigators had not returned GWAS results. Several had experience returning results in the context of linkage/family studies, and many felt that it will become a larger issue in whole-genome/-exome sequencing.

Conclusions—Research context and nature of the study are important considerations in the decision to return results. More nuanced ethical guidelines should take these contextual factors into account.

Keywords

ethics; genome-wide association; genomics; policy; return of results; whole-genome sequencing

The question of returning individual research results and incidental findings has stimulated extensive policy discussion and intense scholarly debate over the past several years. In the context of genome-wide research, it has been identified as one of the most pressing ethical

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challenges warranting immediate policy attention [1]. There is significant disagreement among scholars in the ethics and research communities about the return of research results. Some are hesitant about the return of individual research findings, arguing that individuals should not have access to this information because "preliminary research results are often not replicated, making early findings ambiguous at best" [2]. In addition, many argue that routine return of results, especially in certain research contexts, would impose a significant and unjustified burden on the research enterprise and frustrate the aims of researchers [3]. Others argue that there is a moral obligation to return at least some research results, contending that it upholds the principle of respect for persons and avoids treating research participants merely as a means to an end [4]. Greely argues that, in addition to the need to demonstrate respect for participants, there is a beneficence-based obligation to return at least some individual results. He goes so far as to opine that, at least in extreme circumstances where the results "pose a very high risk of a serious disease", not offering to return results seems "immoral, possibly illegal, and certainly unwise" [5].

Guidelines also suggest there is an obligation to return results of genetic research, at least in some circumstances. Knoppers *et al* analyzed 30 international policies and ethics guidelines related to genetic research and found that "at the international level there may be an emerging ethical 'imperative' to return results in genetic research" [6]. In the USA, the obligation is generally limited to only clinically valid and significant research results [7, 8, 101], rather than a broad right of access to information. Most US guidance documents stress process, which includes the importance of developing a plan for return of results that is approved by an institutional review board (IRB), informing participants of this plan, and offering the option of receiving results to participants during the original informed consent process so as to preserve the right not to know [102,103].

These guidance documents have been criticized for trying to create a one-size-fits-all approach to the return of results. As Beskow and Burke point out [9], context matters. They argue that the degree of vulnerability of research participants (i.e., are they being recruited because they are patients?), depth of relationship between researchers and participants (i.e., is there a longitudinal relationship?) and degree of dependence of the participants (i.e., can participants get access to genetic information elsewhere?) are vital considerations in deciding whether or not to return results. Other important considerations that have received scant attention in the literature are the type of study conducted and the type of technology used.

We recently conducted interviews with investigators of genome-wide association studies (GWAS) about their perspectives and experiences with returning genomic research results to participants. We found that few GWAS investigators have returned results in the context of GWAS; furthermore, many expressed doubt that results will be discovered in the course of GWAS that one would consider returning. However, several investigators had experience returning results in the context of other studies, typically using linkage and family study designs. In addition, many posited that returning results will become a larger issue in the context of whole-genome and whole-exome sequencing. When results are discovered that would warrant return, the nature of the relationship with participants and what was written in the informed consent document seem to be the most important considerations for GWAS investigators in deciding whether or not to communicate those findings.

Materials & methods

We identified GWAS investigators to interview using 'A Catalog of Published Genome-Wide Association Studies', a list generated and maintained by the National Human Genome Research Institute [104]. This list included 360 distinct corresponding authors for 517

published studies. Three were unreachable; the remaining 357 authors were invited to participate in an online survey about their practices and perspectives on returning genetic research results to study participants. Two hundred completed the survey (the results of the online survey will be published separately). At the end of the survey, participants were asked if they were willing to be contacted to participate in a follow-up interview to explore their experiences and attitudes with return of genetic results. If the investigator did not feel he or she was the most appropriate person for the interview, he or she could provide an alternative contact of someone who was familiar with the identifying GWAS. Those who agreed to the interview and individuals named as alternative contacts were sent an email invitation for the interview and were contacted up to three times by email for scheduling. Of the 200 GWAS corresponding authors who took the survey, 73 agreed to be contacted for a follow-up interview or provided an alternative name and contact information. GWAS investigators were interviewed until theme saturation was reached. Saturation occurs when information in the interviews becomes redundant and further interviews add no new information to the analysis [10]. In this study, theme saturation was reached after 35 GWAS investigators were interviewed.

Interviews were conducted over the telephone, verbal consent was obtained, and interviews were audio recorded and transcribed verbatim. Conventional content analysis was conducted [11], using ATLAS.ti™ qualitative software [105]. Investigators were compensated with a US$50 gift card for their participation. All materials and methods were approved by the IRBs at Baylor College of Medicine (TX, USA) and Dana-Farber Cancer Institute (MA, USA).

Results

We report here the results of the interviews with GWAS investigators, responses from whom provide important insights into how the context of the research study impacts policies related to return of results. Investigators interviewed were representative of the sample surveyed and were predominately male (63%), identified as non-Hispanic white (80%), and worked in a university or academic medical center setting (80%) in the USA (60%). The majority (63%) had a PhD, 9 (26%) had an MD, and 4 (11%) had both MDs and PhDs (Table 1). Of those interviewed, 12 (34%) had some experience with returning genetic results, although none in the context of GWAS. 71.4% agreed that genomic research results should be returned under at least some circumstances, and 28.6% agreed that results should never be returned.

Importance of the nature of the study & technology used

Investigators who had experience returning genomic research results to participants did so primarily in the context of linkage or family studies, rather than in the context of the GWAS on the basis of which they were selected for this study. Unlike GWAS, linkage and family studies are designed to identify variants that result in a high risk of disease, and thus are considered more clinically significant. By contrast, GWAS, as described below, have resulted in identification of variants with statistical significance but very small individual impact on disease risk. In addition, in linkage and family studies, investigators or collaborating physicians typically have a longitudinal clinical relationship with the participant and, often, with his or her family members. Investigators who had experience returning results in family or linkage studies but were not the participant's treating physician emphasized the importance of working closely with the physicians interacting directly with the families. Decisions about the return of results were generally made in the initial stages of study design, usually in consultation with the treating physician, and were typically included in the IRB protocol and informed consent document. An investigator with experience returning results explained:

> "We put in to the NIH grant and the IRB protocol from the beginning that if we ever discovered something that we thought would be clinically relevant in our research laboratory, that we would go back to the physician who was responsible for submitting the samples and let them know that there was a discovery that may impact their family and that they should have a discussion, with or without a genetic counselor about potentially submitting samples for a laboratory test where results would be disclosed as part of the normal course of business."
>
> – Subject 283

Those who had returned genetic research results to participants expressed favorable opinions about the experience. For example, one investigator described how returning *BRCA2* mutations found in male prostate cancer participants to additional family members after the participants were deceased proved to be a positive experience:

> "We found that people did want to know, even though it meant that we were actually talking about results about somebody in their family who died … And the other thing we found was that actually a lot of people expressed that it actually helped them with the grieving process because they felt that something positive had come out of their relative taking part in research."
>
> –Subject 168

For these investigators, the potential significance of the findings for the patient and family members and the researcher's relationship with the participant and family members were key considerations when deciding whether or not to include an option to return genomic research results.

Despite this experience, investigators noted that return of results is not the intended purpose of GWAS and doubted whether GWAS would produce any results that would meet criteria for disclosure. Specifically, GWAS results usually demonstrate very small effect sizes that are unlikely to be meaningful to most individual participants. One investigator explained:

> "So, anything that's coming out of a GWAS in a normal sample size study is going to be common, and it's going to have a very, very small effect. And if your risk is [that] there's a 98% chance that you're not going to get the disease, how meaningful is that clinically? Anything coming out of a GWAS, I don't think is going to be relevant to [an individual]."
>
> – Subject 329

As another investigator clearly described, this type of research is not intended to produce individual level results that are clinically meaningful, a fact that some participants may not understand:

> "From past experience, we have found that the issue of returning results in this type of research is problematic. So we prefer we don't get into it at all. [It's problematic] because there are no results. People who participated, there's a gap between expectations and reality. As much as we explain it to participants in studies of this kind, they do not see that in effect there are no results that pertain to them. One cannot say anything about the individual's risk for the disease on the basis of the results that were obtained. These are not studies which look at individual genetic risk, but they calculate overall statistical risk for a population based on a particular sample. And so, there are no results that are referable to the individual."
>
> – Subject 269

Interestingly, investigators did not express concern about the discovery of incidental findings in GWAS because the study design focuses on analysis of aggregate data as opposed to interrogation of individual-level variants. However, many did feel that the issue of returning results, including incidental findings, will likely become a more prominent and pragmatic concern as use of next-generation sequencing technologies proliferates. As one investigator described:

> "Of course the issue [of whether to return] becomes a much bigger one when you plan to move into other types of technologies, for instance whole-exome sequencing, whole-genome sequencing. And I think that this is a very important principal discussion because obviously, what we are looking for and what we are finding and what we are reporting would not be useful to the individuals as for now [with genotyping arrays]. But working with other types of technologies you could envision that there is information in the data on, for instance, breast cancer genes irrelevant to our study aims but, nevertheless, that there could be information in the data that could potentially be of a clinical relevance and, therefore, could be of interest to the patients."
>
> – Subject 238

From GWAS to next-generation sequencing: challenges with return of whole-genome sequencing/whole-exome sequencing results

Considering the transition from GWAS to whole-exome sequencing and whole-genome sequencing, investigators expressed concerns about the implications and impact of returning genomic research results and incidental findings to participants. Concerns were often about the uncertainty of the information to be returned, as well as the impact on scientific progress due to the burden that a duty to warn might impose. Investigators noted how much is still unknown about the massive amounts of data to be generated from sequencing the genome:

> "The next step in research is also to move to high-throughput sequencing analysis where we will start analyzing initially the exome data, and in a later phase the full genome data, and that will uncover all of the genetic variation there is to see in an individual. And I think the big challenge there is to understand about what those variations mean, because the number of variations where we know very little about it or nothing about it is really huge. So I think there's a large gap between identifying those variations in individuals and understanding them. We're really quite far from that."
>
> – Subject 392

Investigators seemed keenly alert to the potential impact on the progress of science, especially when considering whether researchers would be charged with actively looking across the entire genome for relevant findings that warrant return. One investigator explained:

> "If you really are aware of [an incidental finding] and it's there, I would say it's not ethical not to return it. However, then there is the issue should you actively look for it, and I think this is an almost impossible question, because many of the variants that you will find [with whole genome or exome sequencing] are novel."
>
> – Subject 238

Another investigator noted:

> "Let's imagine you did a whole-genome sequencing study and you were interested in some particular genes for schizophrenia but it turned out that your patient had a BRCA1 mutation that was known to be associated with an increased risk of breast

> cancer but had nothing to do with schizophrenia or what your research was. But you have a whole-genome sequence in hand, so it's in there. This is a real problem, but I don't see how it's possible that a schizophrenia researcher could possibly be responsible for scanning the person's genome. There's an infinite number of new findings that come out and mutations and so on, and we're not a major company that's constantly updating its databases and scanning the world literature of all possible disease-causing mutations, and it's unfair to put us in that position."
>
> – Subject 268

There were also concerns that returning genomic research results to participants would further blur the lines between clinical care and research. Some investigators were clear about this distinction; as one explained:

> "We don't do these studies to find, to give clinical guidance to patients. We do these studies to identify biological pathways that may tell us something about pathogenesis, and that's a completely different set of questions. For that reason, we're not trying to generate clinical information, and I don't think it's appropriate to give people half-baked clinical information that we don't really understand."
>
> – Subject 127

Others spoke to the challenge of maintaining this boundary, especially when considering the different researcher–participant relationships. As one investigator described:

> "These are clearly research studies, and I think our obligation is different as a research study. I think if you are the treating physician recruiting patients, you are in an ambiguous situation and you have more of an actionable position because you are a treating physician."
>
> – Subject 119

Ultimately, the decision about whether or not to return results for many investigators came down to what was written in the informed consent document. If the consent form addressed return of results, investigators felt that they were bound by what was written in the informed consent document. As one investigator explained:

> "…there's an obligation to return results if the consent form has stipulated that results will be returned … If the consent form says that results will be returned, you have to return results."
>
> – Subject 269

On the fip side:

> "if the consent says we won't give back information, which is the most expedient kind of consent to write, [then] we won't; won't is won't."
>
> – Subject 392

Discussion

Although some GWAS investigators report having experience with returning results in the context of linkage or family studies and with participants who are also their patients, and many agree that there are some circumstances that warrant returning results, most have not and do not plan to return results of GWAS. This is because investigators viewed the purpose of GWAS to be the analysis of population risk associated with common variants, which typically have very small effect sizes. Thus, they did not perceive it as likely that any results discovered during the course of GWAS would be reportable. Investigators did, however,

think that the issue of returning results would become much more problematic with the proliferation of whole-genome and whole-exome sequencing. This is because genome sequencing is more likely to find mutations with a higher impact on disease (as was true of older linkage studies). Investigators also felt that genome sequencing is more likely to uncover incidental findings that impact disease outside the scope of the original study; interestingly, they did not seem as concerned about the discovery of incidental findings in the context of GWAS.

In the context of whole-genome sequencing and whole-exome sequencing, while researchers recognized that it may be unethical not to return some results or incidental findings, many felt that it was beyond the scope of researchers' duties to interrogate the genome in search of such findings and worried that imposing such a duty would overly burden the research enterprise. McGuire and Clayton have cautioned that establishing such a “duty to hunt” would expand genomic researchers' responsibilities beyond those even of clinicians, and could be used as evidence of what the standard of care should be in genome research, which could lead to potential liability for failure to return [12]. Given the fact that most GWAS investigators report that they are not currently returning results, coupled with the lack of consensus on what results and/or incidental findings should be disclosed [13] and the concern that genomic researchers might quickly become overwhelmed with the number of reportable findings [14,15], policies need to clarify limitations on researchers' responsibilities in this area. They should also address the resource challenges raised by investigators in this study: who is going to pay for the return of results, and is returning results the best use of research dollars?

As several GWAS investigators pointed out, establishing an obligation to return results in genome research fails to recognize a sharp distinction between research and clinical care. Some believe this is unavoidable and argue for a set of clinical obligations to be adopted in this type of research [16]. However, GWAS investigators in this study seem to reject this conflation, especially when the investigator is not also the participant's treating physician. They emphasized the importance of working closely with the treating physician to return results, when deemed appropriate, but they resisted the idea of a general duty to disclose, even in whole-genome sequencing and whole-exome sequencing in which they perceived t he relevance of findings to individual participants to be much greater than in GWAS. This tension deserves additional study and careful consideration as new policies for the return of results are developed.

Finally, GWAS investigators emphasized the importance of what is communicated to participants in the informed consent document. Many saw the consent form as a contract between the researchers and the participant and felt that the researcher was bound by commitments made in the consent document. There was very little emphasis on the informed consent process, and almost no discussion of the quality of information provided in the consent document. Research suggests that study participants often have difficulty understanding basic information described in informed consent documents [17,18], especially as they relate to complex concepts such as genomic research and return of results [19–21]. A more thorough review of consent documents for language about the return of results, and more in-depth study of participants' comprehension and expectations for results in light of what is described in consent forms, would be informative.

Conclusion

Ethical guidelines and policies related to the return of individual genetic research results need to be more context-specific. The type of research conducted and the technology used are important considerations that may influence the decision of whether or not to

communicate results with individual study participants. Guidelines and policies that do not take into account current practices and perspectives of investigators will be ineffective in changing behavior.

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Executive summary

Importance of the nature of the study & technology used

- Individual genetic research results from genome-wide association studies (GWAS) are generally not communicated to study participants.
- GWAS investigators do not perceive the results of GWAS to be reportable because they typically relate to common variants with small effect sizes.
- Some GWAS investigators have experience returning results in other types of studies, typically using linkage and family designs.
- Investigators emphasize the importance of working closely with the participant's treating physician when returning results.

From GWAS to next-generation sequencing

- GWAS investigators believe that the issue of return of results will become much more problematic with whole-genome and whole-exome sequencing because of the potential to discover mutations with higher impact on disease and clinically relevant incidental findings.

Conclusions

- Ethical guidelines and policies should consider the research context, specifically the nature of the study and technology used.
- Additional research is needed on how the issue of returning results is communicated to and understood by research participants during the informed consent process.

Table 1

Sample participant characteristics.

Characteristic	Survey participants (n = 200); n (%)[†]	Interview participants (n = 35); n (%)[†]
Age (years) – mean (SD)	47. 5 (8.7)	45 (9)
Sex		
Male	142 (71.0)	22 (62.9)
Female	58 (29.0)	13 (37.1)
Race/ethnicity		
Hispanic or Latino	5 (2.5)	2 (5.7)
White, non-Hispanic	152 (76.0)	28 (80.0)
Minority, non-Hispanic[‡]	43 (21.5)	5 (14.3)
Degree(s) held		
MD	50 (25.0)	9 (25.7)
PhD	93 (46.5)	22 (62.9)
MD and PhD	42 (21.0)	4 (11. 4)
Other[§]	15 (7. 5)	0 (0.0)
Work setting		
University or academic medical center	164 (82.8)	28 (80.0)
Pharmaceutical or biotechnology	13 (6.6)	2 (5.7)
Government	13 (6.6)	0 (0.0)
Other[¶]	8 (4.0)	5 (14.3)
Location of work setting		
USA	95 (47.5)	21 (60.0)
Europe	71 (35.5)	10 (28.6)
Other[#]	33 (16.5)	4 (11. 4)
Nature of data acquisition		
Primary data/specimen collection	126 (6 3.0)	2 0 (5 7.1)
Secondary data/specimen ana lysis	74 (37.0)	15 (42.9)

[†]Number (%) unless otherwise indicated. In the survey participants column, denominators may not equal 200 due to missing data for individual questions.

[‡]Minority, non-Hispanic includes: Asian, Black or African–American, and other (not specifed).

[§]Degrees held other includes: MD, MPH (n = 4); MD, other (n = 3); other (n = 3); PhD, MPH (n = 2); PhD, other (n = 3).

[¶]Work setting other includes: nonprofit and private research center, institute and/or hospital.

[#]Location of work setting other includes: Canada, Asia, Israel and Australia. SD: Standard deviation.