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“Information is Information”: A public perspective on incidental findings in clinical and research genome-based testing

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Abstract

Background—The potential for genomic incidental findings is increasing with the use of genome-based testing. At the same time approaches to clinical decision making are shifting to shared decision-making models involving both the healthcare community and the public. The public’s voice has been nearly absent in discussions on managing incidental findings.

Methods—We conducted 9 focus groups and 9 interviews (N=63) with a broad cross-section of lay public groups to elucidate public viewpoints on incidental findings that could occur as a result of genome-based testing in clinical and research situations. Data were analyzed using qualitative content analysis.

Results—Participants wanted incidental findings disclosed to them whether or not these were clinical or research findings. Participants used different terms to define and describe incidental findings; they wanted to know that incidental findings are possible and be given a choice to learn about them. Personal utility was an important reason for disclosure, and participants believed that managing information is a shared responsibility between professionals and themselves.

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Conflict of Interest

The authors have no conflict of interest to disclose.

Conclusion—Broad public input is needed in order to understand and incorporate the public's perspective on management of incidental findings as disclosure guidelines and policies are developed in clinical and research settings.

Keywords

genome-based testing; incidental findings; public perspective; personal utility

Introduction

Incidental findings (IFs) have been defined as clinical or research findings that are unanticipated or unrelated given the original reason or purpose for which a person is being tested (1, 2). We are in the midst of a rapid expansion of the use of genome-based screening and testing (1), such as genome chromosomal microarray analysis (CMA), whole-genome sequencing (WGS) and whole-exome sequencing (WES) in both the clinical and research settings (3,4). At the same time, approaches to clinical decision making are shifting to shared decision-making models involving both the healthcare professionals and the public (5-7). The rapid expansion of genome-based tests coupled with the public's expectation to collaborate with their providers in healthcare decision making increases the likelihood not only that IFs will be found during testing (8), but also that the public may want to have a voice in how this information is managed.

Guidelines regarding disclosure of genetic IFs to patients and research participants are based on the collective experiences of clinicians, researchers, and scholars (2,9-12). Ultimately the goal of this collective body of work is to aid in decision making on what to disclose, prioritize returning potentially copious amounts of information, and to protect the public from harm while preserving their autonomy and right to determine what they want to know (13).

The public's voice has been nearly absent in discussions on managing IFs. Several recent studies report that the public favors return of individual research results (IRRs), but the authors do not make a clear distinction between IRRs and IFs (14-19). IRRs, might not be considered IFs depending on a stakeholder's (e.g. patient vs. provider) point of view and the original purpose for the sample collection. Further, the public may feel differently about IFs compared to other IRRs. There are few studies that address the public's views on IFs resulting from clinical genetic or genomic testing. Turney (20) found a lack of consensus among members of the public on preference for disclosure of misattributed paternity identified in *clinical* genetic testing. Townsend et al. (21) found that lay groups wanted a choice to learn about IFs resulting from clinical WGS, and accepted that anxiety and uncertainty might be a consequence of IF disclosure. Hence, the purpose of this qualitative study was to elucidate public viewpoints on IFs that could occur in genome-based testing in clinical and research situations by engaging a broad cross-section of the lay public from diverse backgrounds in focus groups or individual interviews.

Materials and Methods

This study is part of a larger exploratory study examining the management of IFs from the perspectives of numerous stakeholders, including members of the public, medical genetic specialists, researchers, and Institutional Review Board (IRB) chairs (7, 22-25). For this study members of the public were purposely recruited through community-based agencies across a Midwestern state in the USA, including a university-based Center for Aging, a State Department of Public Health, churches, support groups, community groups, community colleges, and a university-based medical genetics department. Flyers and letters about the study were distributed by the community agencies. The study was approved by the University of Iowa Institutional Review Board. Recruitment efforts resulted in nine focus groups and nine individual interviews.

Interview Guide

We developed a three-part interview guide: 1). Word association, 2). Provision of information, and 3). Discussion of vignettes. Details of the interview guide are presented in Table 1. After presenting the vignettes (26, 27) we asked participants a series of open-ended questions (Table 2), changing the nature of the IF to include high risk single gene disorders, increased risk for complex disorders, positive carrier status, disorders that varied in treatment and prevention options, misattributed paternity, and variants of uncertain significance. Participants also completed a socio-demographic survey. All sessions were audio-recorded.

Focus Groups

Focus groups included diverse representation: African Americans, clergy, elders, Hispanics (two groups, one English speaking and the other Spanish speaking), parents of children with hearing impairment, rural community, support groups, and young – middle aged adults. The African American, clergy, rural community, support and young—middle aged adult groups consisted of people who already met regularly (Table 3). Accordingly, we conducted these five focus groups during one of their regularly scheduled meeting times. The remaining four focus groups consisted of individuals who came together for the sole purpose of the research study. The elder and two Hispanic groups were held in a community setting; the parent focus group was held by telephone. Focus groups consisted of two to ten participants and lasted between 60 to 90 minutes. The smallest focus group lasted 60 minutes; all others lasted approximately 90 minutes. The first author conducted the focus groups. A second member of the research team observed and recorded field notes on non-verbal communication patterns. The Spanish-speaking Hispanic focus group was conducted in Spanish with a bilingual interpreter whose native language is Spanish.

Individual Interviews

Individual telephone interviews were conducted by the first author with parents whose children had a chromosomal microarray analysis (CMA) because they were not available to meet in a central location. The parents were identified through a university-based medical genetics department. Finally, an individual interview was conducted with one member of the

young – middle aged group and one parent whose child has a hearing impairment. These individuals were unable to attend a focus group. Interviews lasted between 30 to 60 minutes.

Data Analysis

Each transcript was transcribed verbatim, and checked for accuracy. To verify the interpretation that took place during the Spanish-speaking focus group we used a double translation process (28) providing two Spanish to English translations, one in real time (interpretation) and one after transcription (translation). We found no coding discrepancies when the two were compared.

We used conventional content analysis (29). All transcripts were independently coded by two members of the research team (SDH and AH) who met weekly to discuss coding and resolve any discrepancies. The codes were organized into categories and subcategories using the NVivo 8 software program (30) to facilitate data management and analysis. A third research team member resolved discrepancies in coding.

Results

Sixty-three participants were recruited into the study. Their socio-demographic characteristics are summarized in Table 3. The total sample is divided into nine groups for the purpose of reporting the data and because the participants were purposely recruited to represent a specific demographic characteristic.

Findings

Data were classified into three main categories: 1) What is an IF, 2) Knowing about IFs, and 3) Managing IF information. Findings are summarized by categories and subcategories in Table 4 and presented in more detail below. Participants wanted incidental findings disclosed to them whether or not these were clinical or research findings. Unless specified, the selected quotes represent pervasive attitudes within and across groups.

What is an IF

Participants largely viewed IFs as unexpected findings of “bad news”. Results could be unexpected by both the patient and the clinician — as this quote illustrates:

The nurse explained to me that he didn’t have the deletion they anticipated, they had found something [else] that they didn’t expect to find, that he has an extra X chromosome.

Or the result could be unexpected by the patient, but plausible to a clinician:

When they came in my room and told me the results of the hearing screen were that my child couldn’t hear. I felt like the results were completely unexpected.

Other descriptors for IFs are found in Table 4.

Knowing about IFs

We Want To Know—Participants primarily wanted an IF disclosed to them, regardless of the severity of the associated condition, certainty of its meaning, or context for the test (clinical or research): I don't see a difference between a clinical and research setting; information is information. Prevention, treatment, and examples of personal utility were reported in every group as possible reasons for wanting IFs disclosed to them (Table 5).

Participants also wanted to know about variants of uncertain significance (VUS), although they were less enthusiastic about VUS compared to IFs with some known information about its significance. VUS were viewed as something an individual, healthcare provider or researcher should document and monitor least they become important later:

(VUS) is something that I would want put into a letter or some type of document like that. Have it sent to my doctor and have it on file in case something does happen or a clinical study and they find that... maybe it will be significant in 5 years.

Others May Not Want To Know—Only three participants (three different groups) explicitly said that they did not want to know about IFs stating that there were other ways to identify their risk, that worry might affect their quality of life, or that they feared the information might affect insurability.

While the prospect of worry was acknowledged, it did not appear to be an overwhelming reason to keep participants in this study from wanting to know about IFs. Rather, worry was a manageable trade-off for the benefits of learning about IFs:

(The IF) is another thing to worry about, but I think that worry kind of edges away the more prepared you are for the situation.

The possibility that providers or researchers might withhold IFs came up indirectly and spontaneously. Participants from the clergy, African American, rural community, and English speaking Hispanic groups shared personal stories about health information being withheld from them, a family member, or an acquaintance affirming that withholding information to keep people from worrying is no longer acceptable, "I can't imagine a charade like that going on today."

How We Want To Be Told—Participants emphasized the need for interventions such as patient education or a consent process prior to testing that specifically prepares them for the possibility of an IF and gives them a choice to learn about different types of IFs.

Choices, people like choices. Having a survey or a check box thing is a good way to please everybody. Then they feel like they're in control of their own health and their own decisions instead of having the doctor or nurse (disclose information you don't want to have).

Participants' opinions varied within groups on who the best person would be to disclose IFs (e.g. personal physician, specialty physician, or researcher). However, participants across all groups consistently described a preference for receiving information from someone they

trusted and someone who was either knowledgeable about the finding itself or who would make a referral to a knowledgeable specialist.

Participants repeatedly suggested involving a personal physician or a healthcare specialist with whom people had an ongoing relationship. However, it was the quality of the relationship not the person that was emphasized. For example, when an IF is derived from research or clinical testing and the researcher or clinician did not have an ongoing relationship with the participant, then involving a professional who did was valued. That person was viewed as someone who could personalize the interpretation of the result beyond its biological meaning as illustrated in this dialogue:

P1 Whereas a researcher could give you the information,

P2: He may not be able to interpret it...

P1: yes, not because the researcher is not any less skilled or anything just because the context. It may be harder to communicate in a holistic way, what this means to you or what it might mean to you the researcher does not know you.

On the other hand, IFs resulting from research were viewed as potentially more private than IF's resulting from clinical testing, and thus grounds for not involving a clinician.

I don't think it's the researcher's job to figure out the repercussions. The researcher's job is just to inform you and then you decide if you're going back to your other physician and say "tell me what this means in terms of my own life."

Participants wanted IF-related information to come with a plan. They valued guidance from the clinician or researcher about what to do next, where or who they should see next, and in terms of research where they would go to follow up or confirm the research finding.

P:...are they just (going to) spit it out and it's, appointment over? Or, is there (going to be) follow through? Is there (going to be) some (information) regarding this risk, or potential (risk). Like she said, okay, what precautions can I take?

Moderator: So it's not just getting information, it's, do you have a plan?

P: There you go that's what I'm saying...

Managing IF Information

Taking personal responsibility for the management of IFs emerged as an overarching theme across all groups. Participants viewed the management of IF information as a shared endeavor between professionals and lay public. Professionals were viewed as having an obligation to disclose information:

I'm looking big picture, the doctor or researcher ethically and morally has an obligation to tell me, even though they can't figure out what it is, or it's never caused a problem, or they don't know...

However, the ultimate responsibility to act on the information resided with the recipient:

(Information about IFs) is not so different from some of the advice that you would get from your personal physician. She'll often tell you, you need to do this or

you're going to face such and such a result. And she doesn't call you up all the time and say... 'Oh, are you doing your exercises?' You have some assumption of responsibility...

Discussion

Participants used different terms to define and describe incidental findings. They wanted to know that IFs are possible and be given a choice to learn about them, personal utility was an important reason for disclosure, and they believed that managing IF information is a shared responsibility between professionals and themselves.

It is interesting that participants in this study described IFs as findings that are *unexpected to them* regardless of the plausibility of a result. An important first step to discussing IFs with patients and research participants is for the professional (clinician or researcher) and patient or research participant to come to a mutual understanding of what constitutes an IF in each testing situation. This ought to be done up front so that discussions can progress to making decisions about whether one desires IF disclosure. What exactly constitutes an IF is not consistently defined by clinical and research professionals (11, 22-24) and this first step is an opportunity to clarify language and understanding.

Our findings add to the growing body of evidence that the public would like the option to have IRR returned to them (14-19), and in our study, the IRR was an IF. Clinical genetics specialists' decision to disclose IFs may depend on the provider's definition of the IF, its clinical significance, and assessment of the patient's readiness to receive the information (22) and actionability (31). Berg, et al. (9) proposed a binning system to help clinicians prioritize and determine what type of genetic IFs to disclose to patients. Our findings are consistent with Townsend et al. (21) and show that members of the lay public want to be given the choice to learn about IFs in clinical genome-based testing, and in our study, this was not dependent on nature of the IF. We would add that disclosure of IFs in both clinical and research context has the potential to inform and enrich dialogue between professionals and the public, and might lead to more convergence on attitudes toward disclosure of IFs (32). Impeding the public's access to their information limits their role in healthcare decisions (5, 6, 33). Further, disclosure provides opportunities to inform members of the public about genetics and genomics within a personally relevant context (34).

Scholars emphasize clinical utility as a priority for disclosing IF information in their disclosure guidelines (2, 9-11). Some scholars also acknowledge that genetic information may be of personal benefit for a patient or research participant regardless of its clinical utility (8, 35). For example, Fabsitz et al. (36) and Wolf et al. (11) proposed that the personal value a patient or research participant places on specific information should be a deciding factor for what might be disclosed, especially when evidence of clear clinical utility is questionable. Members of the public in our study shared this point of view. They embraced both clinical and personal utility and factored these in when considering whether or not they might personally want to know about IFs. At the same time they recognized that others might hold different values with respect to utility and make different decisions, thus further emphasizing their desire to be given a choice to learn about IFs.

One of the most noteworthy findings from this study was the notion of “sharing responsibility” for the management of IF information. Participants viewed management as a shared endeavor with the professional obligated to disclose IFs along with options for follow up, and the recipient taking responsibility for his or her choices and monitoring information despite the fact that it may cause anxiety. Our findings on responsibility are consistent with other lay public’s perspectives reported by Townsend et al. (21). Sharing responsibility for managing IF information is also consistent with shared decision making involving both the medical community and the public (5, 6) and donor-driven enquire process described by Kollek and Petersen (37) and for the disclosure of IRRs in genomic research.

We presented the clinical vignette first followed by the research vignette. This order may have encouraged therapeutic misconception (38) among participants and may account for the similarities in public perception about IFs in clinical and research contexts. However, the fact that participants viewed clinical and research IFs similarly may be indicative of their desire for a more active role in decision making and responsibility for the management of IF information (5,6, 38,39).

A limitation of the study is that few underrepresented minority men participated. We must also consider bias of ascertainment. Members of the lay public may have been motivated to volunteer because they already actively participate and share responsibility for their healthcare decision making. Finally, the use of hypothetical situations does not predict actual behavior. However, the intent of this study was not to predict behavior, rather it was to elucidate public viewpoints on IFs.

This is one of the few studies that actively engaged members of the lay public from diverse backgrounds on the topic of IFs related to genome-based testing. Inclusion of their voice is critical to developing policy and guidelines for best practice surrounding disclosure, whether in a clinical or research setting. Broader public input is needed in order to understand and incorporate the public’s perspective on management of incidental findings.

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Table 1**Interview guide**

Interview guide	Content	Purpose
Part one: Word association on the broad topic of incidental findings.	Each session began by asking, “What comes to mind when you hear the words ‘incidental finding?’”	Generate participants’ descriptions of what constitutes an IF.
Part two: Provision of information	<p>Topics:</p> <ul style="list-style-type: none"> Genetic test Genome-based test Variants of uncertain significance: <ul style="list-style-type: none"> Reliability and certainty of genetics and genomic testing Definition of IF for the purpose of the study: <p><i>-Incidental findings are test results or findings unrelated to the reason or purpose for which a person is being tested.</i></p>	Provision of information in order to engage participants in and open up discussion.
Part 3: Discussion of vignettes Participants were presented with two vignettes [24, 25]; the first was a clinical testing scenario and the second a research scenario.	<p>Clinical vignette: Suppose your daughter is having problems that her doctor thinks might be autism. She orders a genomic test. Results from the genomic test revealed that your daughter has two deletions in her genome. One is likely causative for autism. The other is related to an increase risk to develop [condition]. Results also revealed that you have second deletion.</p> <p>Research vignette: Suppose that you are part of a research study to find changes in genes that cause high cholesterol. Suppose that you have high cholesterol as do many of your family members. Results (incidentally) reveal changes in a gene that is connected with an increase risk for developing [condition].</p>	Situate the larger discussion on managing IFs

Table 2

Discussion questions following the presentation of vignettes

Discussion questions	
<ul style="list-style-type: none">• If you were in this situation what would you want to know?• What reasons do you have for knowing or not knowing?• Describe situations when you would not want to know about an IF?• What information would be important to you to have in deciding if you wanted to be told about the incidental findings related to a genomic testing?• Who should tell you about the IF?	

Table 3

Socio-demographic characteristics by group (N=63)

Group Name (n)	Type of Interview	Community Source	Gender	Age Range	Race/Ethnicity n(%)	Education n (%)
African Americans* (n=8)	Focus group	Local church Bible study group	Male 0 Female 8 (100)	26-61	Non-Hispanic Black 8 (100)	<High School 0 High School 3 (37.5) 2 Year Degree 0 4 Year Degree 1 (12.5) >4 Year Degree 4 (50)
Clergy* (n=4)	Focus group	Community ecumenical committee	Male 2 (50) Female 2 (50)	57-60	Non-Hispanic White 3 (75) American Indian 1 (25)	<High School 0 High School 0 2 Year Degree 0 4 Year Degree 0 >4 Year Degree 4 (100)
Elders (n=6)	Focus group	University center for aging	Male 2 (33) Female 4 (67)	67-82	Non-Hispanic White 6 (100)	<High School 0 High School 0 2 Year Degree 0 4 Year Degree 2 (33) >4 Year Degree 4 (67)
Hispanic (n=7)	Two focus groups Spanish speaking (n=5) English speaking (n=2)	Community based clinic	Male 0 Female 7 (100)	26-46	Hispanic White 5 (71) American Indian 2 (29)	<High School 2 (29) High School 5 (71) 2 Year Degree 0 4 Year Degree 0 >4 Year Degree 0
Parents of hearing impaired children (n=3)	One focus group (n=2) One individual interview	State department of public health	Male 0 Female 3 (100)	26-39	Non-Hispanic White 3 (100)	<High School 0 High School 0 2 Year Degree 0 4 Year Degree 1 (33) >4 Year Degree 2 (67)
Parents whose children had CMA testing (CMA) (n=7)*	Individual interviews	University medical genetics department	Male 1 (14) Female 6 (86)	31-51	Non-Hispanic White 6 (86)	<High School 0 High School 4 (57) 2 Year Degree 1 (14) 4 Year Degree 1 (14) >4 Year Degree 0
Rural community* (n=10)	Focus group	Volunteer fire department	Male 4 (40) Female 6 (60)	26-57	Non-Hispanic White 10 (100)	<High School 0 High School 1 (10) 2 Year Degree 8 (80) 4 Year Degree 1 (10) >4 Year Degree 0
Support group* (n=9)	Focus group	Recovery support group	Male 3 (33) Female 6 (67)	21-60	Non-Hispanic White 6 (56) Non-Hispanic Black 2 (22) American Indian 2 (22)	<High School 0 High School 5 (56) 2 Year Degree 1 (11) 4 Year Degree 3 (33) >4 Year Degree 0
Young - middle aged adults* (n=9)	One focus group (n=8) One individual interview	Community college Emergency Medical Technician Class	Male 6 (67) Female 3 (33)	21-49	Non-Hispanic White 8 (89) Hispanic White	<High School 0 High School 3 (33) 2 Year Degree 3 (33)

Group Name (n)	Type of Interview	Community Source	Gender	Age Range	Race/Ethnicity n(%)	Education n (%)
					1(11)	4 Year Degree 2 (22) >4 Year Degree 1 (11)

* Groups consisted of people who already met regularly

** one person did not report education level

Table 4

Summary of findings by main and sub-topical categories.

Main category I	What is an IF
Subcategories	<ul style="list-style-type: none"> -Primarily unexpected results -Other descriptions of IFs <ul style="list-style-type: none"> • Accidentally found • anticipating a test to yield a specific result and unexpectedly receiving a different one • Unrelated to the original purpose of the test • Excessive information • Not important to your health
Main category II	Knowing about IFs
Subcategories	<ul style="list-style-type: none"> -We want to know <ul style="list-style-type: none"> • Prevention • Treatment • Personal utility -Others may not want to know -How we want to be told <ul style="list-style-type: none"> • Be prepared for the possibility of IFs • Be given the choice to learn about IFs • Personalized disclosure by knowledgeable provider • Disclosure should come with a plan
Main category III.	Managing IF information
Subcategory	<ul style="list-style-type: none"> -We share responsibility <ul style="list-style-type: none"> • Obligation of the professional to disclose Ifs • Personal responsibility to act or not act upon the information

Table 5**Exemplar Quotes of rationale for disclosure of IFs**

Response category	Exemplar quote
Prevention	<i>If you can prevent something, then of course you would want to deal with that situation now rather than later.</i>
<i>Treatment and early intervention</i>	<i>P1*: ...even if it was they were testing for breast cancer and the autism gene came up. I think that is a necessary disclosure too because of early intervention. P2: Well you might not be able to prevent (a disease), but you might be able to work on minimizing the effects of it—slow it down.</i>
Personal Utility**	
➤ Life plans—how you live your life	<i>I guess, still, it (a variant associate with an increase in risk) is a life altering thing even though something is not definite it still changes the way you live your life ...because if you tell me I might have a 50% chance of having cancer, but if I eat carrots every day for the rest of my life... that changes my life.</i>
• Retirement	<i>If genetics could predict when we're going to die, that would definitely affect when people choose to retire (laughter).</i>
• Career/school plans	<i>If (your child) wanted to go to school and they wanted to wait to do it and you know (they are at risk to develop a disease), then they might end up not having that chance.</i>
• Reproductive plans	<i>...can also be very helpful planning for a family. (another person adds: right, right)</i>
➤ Change the priorities in your life	<i>P1: Spending more time with your family and changing your priorities. Group: Yeah. P2... maybe like that song live like you were dyin' P3: ...look at life differently, not take it for granted</i>
➤ Get your affairs in order	<i>(Y)ou can have all your affairs in order, like you can make sure the car titles are assigned to your mate or your loved one, you can have a will ready, plan a funeral ahead of time. You can just have all that kind of stuff ready.</i>
➤ Explanation for why things happen	<i>More than anything just to understand why things are or might be the way they are...this way you're aware and well informed.</i>

* P= participant. A number following the P signifies multiple participants input to the specific quote.

** For this study personal utility is defined as benefits or harms that are manifested outside of medical context and may indirectly affect medical outcomes (32).