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Provider Perspectives

Genetics specialists' perspectives on disclosure of genomic incidental findings in the clinical setting☆

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ABSTRACT

Objective: Evidence documenting management of incidental findings (IFs) from clinical genomic testing is limited. The aim of this study was to examine genetics specialists' perspectives regarding current and preferred disclosure of clinical genomic IFs.
Methods: 50 genetics specialists, including medical geneticists, laboratory professionals, genetic counselors, and nurses participated in structured telephone interviews. Data were analyzed using qualitative content analysis and descriptive statistics.
Results: Most specialists had encountered IFs, but definitions of IFs varied. They discussed challenges with informing patients about the prospect of IFs and disclosing IFs to patients. Causing psychological harm to patients was a concern. Participants were divided on whether IFs needed to be clinically significant and/or actionable in order to be disclosed to patients. Creating formal disclosure guidelines was considered useful, but only if they were flexible. Additional counseling, more interdisciplinary communication, maintaining contact with patients, and a centralized database to interpret IFs were also proposed.
Conclusion: Genetics specialists offer insights into the challenges of defining IFs, knowing when and how to disclose them, and the potential need for flexible disclosure guidelines.
Practice implications: Further discussion between practicing genetics specialists is needed to develop consensus on the development of best-practice guidelines for IF management.

1. Introduction

The rapidly increasing number and use of genetic and genomic tests in clinical practice are raising new dilemmas with respect to management of incidental findings (IFs). Sequencing technologies, including whole genome sequencing (WGS) and whole exome sequencing (WES) increase the possibility of encountering variants known to cause disease, suspected to cause disease, or of uncertain significance that are outside the original intent of testing [1]. Although WGS and WES can reveal findings beyond those related to the purpose of the test, they may be unanticipated and thus still considered IFs [2].

Ethical management of IFs is debated in the research setting [3]. Some members of the public say that they would like to receive individual research results from genomic research [4,5], which may include IFs [4]. While some issues related to disclosure of IFs in research are similar to those in the clinical setting [3,6,7], clinicians typically have a more personal relationship with their patients than researchers, and the clinician role may extend to the duty to warn patients' family members about future health risks indicated by genomic IFs [8].

Genomic IFs may have direct clinical implications for patients and their families' health, have personal utility, be useful for future reproductive decisions or for life planning, or be of personal interest [9]. While the discovery of IFs is a component of clinical practice, the amount of data that can potentially be generated from genomic testing creates new challenges [10].

1.1. Genetic and genomic tests and IFs

While genetics specialists are likely to have some experience managing IFs, the increased volume of IFs due to the increase in the number of tests and genome-scanning technology may mean that more time will be devoted to validating, interpreting, and

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communicating IFs to patients. Furthermore, primary care physicians may ultimately be responsible for implementing follow-up procedures with respect to IFs that enter into patient records [11]. Ethical issues that arise when entire or extensive segments of a patient's genome are interrogated include the risk of providing patients with incomplete or incorrect information; providing information for which patients are not prepared; exposing patients to unnecessary and potentially harmful or ineffective treatment; and determining whether or not to report misattributed paternity, consanguinity, or carrier status [1,12–14].

There is no consensus regarding how to minimize these risks in the clinical setting. One proposal recommends limiting disclosure to IFs with clinical utility, although patients and clinicians may agree to disclose IFs without clinical utility [15]. The proponents of this proposal argue that limiting disclosure to IFs with clinical utility reduces the potential for reporting false positive findings [16] or overwhelming patients and clinicians with currently uninterpretable information [15]. Other recommendations range from offering menu-type options on informed consent documents [17], to a 'blanket' disclosure policy to return all genomic findings, regardless of their significance [12]. Associated issues include whether written informed consent should be required that addresses both the possibility of IFs and whether they will be disclosed to patients and/or family [18].

To our knowledge, there is no empiric research regarding genetic specialists' perspectives concerning disclosure of IFs from clinical genetic or genomic testing. The purpose of this study was to examine the perspectives of clinical genetics specialists regarding the management of IFs.

2. Methods

2.1. Approach

This paper presents one component of a larger study examining the management of genomic IFs from the perspectives of numerous stakeholders (Williams and Simon, NHGRI RC1 HG005786). This report focuses on the perspectives of genetics specialists in the clinical setting. This includes medical geneticists, laboratory professionals, genetic counselors, and genetics nurses. The Institutional Review Boards at The University of Iowa and The University of Northern Iowa approved this study.

2.2. Participant selection

We used purposeful sampling [19] to identify genetics specialists involved in clinical genomic testing. Participants were invited through collaboration with the Heartland Regional Genetics and Newborn Screening Collaborative, the American College of Medical Genetics, the National Society of Genetic Counselors, and the International Society of Genetics Nurses. Potential participants were directed to contact the University of Northern Iowa Center for Social and Behavioral Research (UNICSBR) who screened them for eligibility.

2.3. Interview guide development and pilot interviews

The interview guide (Table 1) was developed by the research team following an extensive review of the literature on the issues related to IFs in clinical practice and research contexts [7,10,20,21] and consultation with clinical experts, including a medical geneticist and a genetic counselor. For this study, we defined IFs as "test results unrelated to the reason or purpose for which a person is being tested; sometimes the health significance of IFs is known, but often their significance is ambiguous" [10,22]. This definition was provided to participants if they asked for clarification of the use of this term in the study. Questions were refined in a one day workshop with PIs, interviewers, and survey methodologists from UNICSBR.

The interview guide was piloted in three phases. In the first phase, members of the research team interviewed a medical geneticist and a genetic counselor with clinical experience in genetic and genomic testing. In the second phase, three members of the research team took the role of genetic specialists in interviews conducted by the UNICSBR interviewers. Minor wording changes were made as a result of these first two piloting phases. The third phase involved the administration of the interview by a UNICSBR interviewer to a medical geneticist and a laboratory director who contacted the interviewing center to participate in the study. No changes were identified upon completion of this last component of the pilot process.

2.4. Data collection and management

Trained interviewers conducted telephone interviews with participants who met eligibility criteria. Interviews were

Table 1
Structured interview guide questions.

1.	When you hear the words, "incidental finding," what comes to mind?
2.	Can you give me an example of an incidental finding that has occurred in your work?
3.	What information about incidental findings, if any, are you able to provide your patients before they undergo a genetic or genomic test?
4.	In what format do you provide this information to your patients?
5.	Apart from any of the information that is already shared with your patients, what additional information about incidental findings do you think should ideally be shared with them?
6.	How do you feel about giving your patients the option of indicating whether or not they want to be contacted if an incidental finding is found?
7.	Next, I would like to get an idea of how many IFs you encounter in your work. Thinking back over the last 12 months, how many genetic or genomic IFs have you encountered?
8.	Is this number more or less typical of most years?
9.	Approximately what percentage of these IFs did/do you contact the patient about?
10.	How much of the detailed information about an incidental finding do you provide to a typical patient?
11.	How did/do you contact (or, "are you likely to contact" for respondents who have not yet done so) a patient with the news that an incidental finding has been discovered?
12.	Do you have specific procedures in place for dealing with or managing incidental findings? If not, would you find such procedures useful in any way? Why? Why not?
13.	Who developed these procedures?
14.	Whose policies or guidelines, if any, are these procedures based on?
15.	What do these procedures require you to do?
16.	How well have these procedures worked for you so far? [*If respondent has not needed to use the plan yet*] How well do you think they are likely to work for you?
17.	Have you personally discussed with patients any incidental findings that you have found?
18.	Approximately how many such discussions have you had over the last 12 months?
19.	How well do you think patients understand the information you typically share with them?