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ORIGINAL ARTICLE

The disclosure of incidental genomic findings: an "ethically important moment" in pediatric research and practice

Martha Driessnack · Sandra Daack-Hirsch · Nancy Downing · Alyson Hanish · Lisa L. Shah · Mohammed Alasagheirin · Christian M. Simon · Janet K. Williams

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Abstract Although there are numerous position papers on the issues and challenges surrounding disclosure of incidental genomic findings involving children, there is very little research. To fill this gap, the purpose of this study was to explore the perspectives of multiple professional (N=103) and public (N=63) stakeholders using both interviews and focus groups. Using qualitative analysis, we identified one overarching theme, "It's hard for us; it's hard for them," and three subthemes/questions: "What to disclose?," "Who gets the information?," and "What happens later?" Perspectives differed between professional (Institutional Review Board chairs, clinicians, and researchers) and public stakeholders. While professionals focused on the complexities of what to disclose, the lay public stated that parents should have all information laid out for them. Professionals pondered multiple parent and child situations, while the public identified parents as informational gatekeepers who know their children best. Professionals described the potential requirement for follow-up over time as a logistical "nightmare," while the public believed that parents have the responsibility for managing their children's health information over time. However, the parent role as gatekeeper was seen as time limited and in need of professional support and backup. Our findings present a case for needed dialogue around what we propose as an "ethically important moment," with the goal of protecting and respecting the viewpoints of all stakeholders when policies regarding children are developed.

Keywords Incidental findings · Genomics · Children · Gatekeepers

M. Driessnack (✉)
Oregon Health & Science University, School of Nursing,
3455 SW US Veterans Hospital Road, Portland, OR, USA
e-mail: driessna@ohsu.edu

S. Daack-Hirsch · N. Downing · A. Hanish · L. L. Shah · J. K. Williams
College of Nursing, The University of Iowa, 50 Newton Road,
Iowa City, IA, USA

M. Alasagheirin
Department of Nursing, Bradley University, Peoria, IL, USA

C. M. Simon
Carver College of Medicine, The University of Iowa,
Iowa City, IA, USA

Introduction

The primary focus of the US National Heart, Lung, and Blood Institute initiative, *No More Hand-Me-Down Research*, is the current need to increase the numbers of children in research. Others have expanded on this initiative, highlighting the need for more *genomic* research involving children. For example, Kohane (2011) argues that children should not only be included in studies to understand the genomic basis of disease but also be *preferentially* studied over adults. Kohane's provocative argument emphasizes that children have distinct physiology and disease risks, and accordingly, there could be no substitute for including them in genomics studies. He states that (1) many forms of adult disease have genetic etiologies that were discovered in pediatric studies, where more penetrant or homozygous mutations presenting in early childhood made the genetic pathogenesis much clearer than in adult disease; (2) being able to detect genetic antecedents of adult disease facilitates our understanding of how disease develops, providing lead time for intervention and/or prevention; and (3) variation from environmental exposures is smaller in children when compared with adults, and therefore, a case could be made that a greater fraction of pathophysiologic variability in children can be attributable to the inherited traits (2011).

The use of genome-based tests, such as chromosomal microarray (CMA), genome-wide association studies, as well as whole-genome sequencing (WGS) and whole-exome sequencing (WES), is also increasing in clinical practice (Brunham and Hayden 2012; Feero et al. 2010; Green and Guyer 2011; Kohane et al. 2006). As one example, the American Academy of Pediatrics and the American College of Medical Genetics now recommend CMA as a first-tier clinical diagnostic test for children with suspected autism spectrum disorder, developmental delay, intellectual disability, and congenital anomalies (Manning and Hudgins 2010; Miller et al. 2010; Shen et al. 2010).

This inclusion of more children in genomic research and the increase in genome-based testing in clinical practice create new challenges for pediatric health-care researchers and practitioners, especially as genomic findings and interpretations become increasingly nuanced (Hens et al. 2011; Kohane 2011; Samual et al. 2012). At the same time, these challenges provide the opportunity for thoughtful engagement, reflection, and response across stakeholder communities (Bush and Rothenberg 2012; Driessnack and Gallo 2011; Hudson 2011; Lantos et al. 2011).

One such challenge and opportunity is the emergence of issues surrounding the identification and reporting of incidental genomic findings in pediatric research and/or practice (e.g., Lantos et al. 2011; Wilfond and Carpenter 2008). Incidental genomic findings can be defined as unsolicited genomic information gained through testing and therefore unrelated, or incidental, to the original intent of the research query or clinical screening (Wolf et al. 2008), which can include, as examples, known mutations, variants of unknown significance, and misattributed paternity or even unsuspected consanguinity (Zawati et al. 2011). While incidental findings, in general, are not new to pediatric researchers or practitioners, with CMA, WGS, and WES now central to research and on the rise in routine clinical care, the discovery of incidental genomic findings is expected to increase (Bick and Dimmock 2011; Cho 2008; Zawati et al. 2011). Once discovered, the challenge then becomes one of deciding if, how, and when to disclose them (Ali-Khan et al. 2009; Avard et al. 2011; Biesecker 2012; Bush and Rothenberg 2012).

A number of moral and pragmatic justifications that support the decision to return or not to return research results have been published (Lemke et al. 2012). For example, some scholars argue that a researcher's primary duty is to conduct research and that this duty should not be hampered by policies requiring the return of individual research results or incidental findings (Hens et al. 2011). Others urge researchers to ensure that any innovations in and through research be equaled by innovative policies that engage, respect, and protect research participants and patients (Hudson 2011). Still others debate whether any "no return" policy is acceptable in any situation (Zawati et al. 2011). In clinical practice, there is a parallel trend, signaling wider adoption of transparency and *shared* decision making in patient care (Coulter 1997; Delbanco et al. 2010; Elwyn et al. 2012; Emmanuel and Emmanuel 1992; Kon 2010; Legare et al. 2010). The goal in shared decision making, which is the process by which health care decisions are made jointly by the practitioner and the patient/family, is to make decisions in a manner consistent with the patient's/family's wishes. This patient-driven process is seen as the crux of patient-centered care (Legare et al. 2010), which has been highlighted by the Institute of Medicine as the central factor related to improving quality and reducing cost of care (IOM 2012). According to the IOM, enacting person-centered care will require the establishment of new approaches that focus on a person's priorities and needs, rather than simply on how the larger research or clinical setting define its own needs, priorities, and outcomes.

To date, there have also been a number of position papers highlighting potential issues and challenges specific to the identification, definition, and disclosure of incidental genomic findings (e.g., Avard et al. 2011; Fabsitz et al. 2010; Hens et al. 2011; Wilfond and Carpenter 2008; Wolf et al. 2012; Zawati et al. 2011). However, there are very few *research* studies. More importantly, there are very few research studies that engage multiple stakeholders, such as parents and advocacy groups as well as clinicians, geneticists, and ethicists, on the unique issues and challenges when incidental findings involve children (Baret and Godard 2011; Reiff et al. 2012; Townsend et al. 2012). Our aim in this paper is to continue to address these gaps.

Methods

We present one piece of a larger exploratory, mixed methods study that focused on issues related to the management of incidental genomic findings in both research and clinical settings (Downing et al. 2013; Simon et al. 2011; Williams et al. 2012). The purpose of the current study was to capture the unique issues and challenges surrounding the discovery and disclosure of incidental genomic findings when the individual involved is a child. Insight into these issues and challenges is important to the development of sound policies and best practice. These data have not been reported in our other manuscripts.

Sample

Purposeful, stratified sampling was used to obtain a broad cross-section of key stakeholders (N=166). The resultant sample included 103 professionals and 63 lay public members. The professionals included 34 Institutional Review Board (IRB) chairs, 19 genetic researchers, 17 genetics nurses/counselors, and 33 clinical/laboratory geneticists. The lay public groups engaged older adults, young adults, rural dwellers, community-based support groups, clergy, African Americans, English- and Spanish-speaking Hispanics, and parents whose

children either had CMA testing or congenital hearing loss identified through newborn hearing screening. Further details are provided in Table 1.

Data collection

Data were collected using semi-structured interviews and focus groups. Interview and focus group guides were developed a priori, following an extensive literature review (Downing et al. 2013; Simon et al. 2011; Williams et al. 2012). They were reviewed by the entire research team for relevance, accuracy, and cross-referencing capabilities and then piloted. All interactions were audio recorded, transcribed verbatim, checked for accuracy, and then entered into NVivo 8 (QSR 2008) to facilitate data management. Phone interviews with IRB chairs, clinicians, and researchers were conducted by trained interviewers from the University of Northern Iowa Center for Social and Behavioral Research. Lay public phone interviews and on-site focus groups were conducted by a member of the research team, who is an experienced focus group leader, genetic specialist, and qualitative researcher from the University of Iowa. IRB approval was obtained from both the University of Iowa and the University of Northern Iowa.

To orient lay public participants to what is meant by incidental findings, participants were invited to share their ideas about incidental findings in day-to-day life and their specific experiences with genetic testing. The leader also provided an explanation for the differences between genetic testing (targeted to examine one gene) and genomic testing (examines substantial amounts of an individual's genome) and any needed clarification. Then, two case study genomic testing vignettes (one clinical and the other research) were presented changing the nature of the incidental finding (e.g., high-risk single gene, positive carrier status, misattributed paternity, etc.) as a method to engage the lay public participants to reflect, discuss, question, and think about incidental findings. This approach is described in more detail elsewhere (Daack-Hirsch et al. 2012, in review). Similar approaches to deliberative engagement of the lay public have been used in other studies exploring the incorporation of public perspectives (e.g., Lemke et al. 2012).

Table 1 Sample demographics

	Professional stakeholders	Public stakeholders	Overall
N (%)	103 (62.0)	63 (38.0)	166 (100)
Sex, *n* (%)			
Males	58 (56.3)	18 (28.6)	76 (45.8)
Females	45 (43.7)	45 (71.4)	90 (54.2)
Age			
Mean	51	41	–
Range	26–75	21–82	21–82
Ethnicity, *n* (%)			
Hispanic	2 (1.9)	8 (12.7)	10 (6.0)
Non-Hispanic	101 (98.1)	53 (84.1)	154 (92.8)
Not reported	0	2 (3.2)	2 (1.2)
Race, *n* (%)			
White	89 (86.4)	45 (71.4)	134 (80.7)
Non-White	12 (11.7)	16 (25.4)	28 (16.9)
Not reported	2 (1.9)	2 (3.2)	4 (2.4)

Data analysis

Using qualitative content analysis (Elo and Kyngas 2008), data from the larger study were initially reviewed line by line and coded into categories. Data assigned to the child(ren) category were then pulled for further analysis, which involved looking within and then across data for recurring patterns and themes (Ayres et al. 2003). The resulting themes were then summarized and organized into a thematic matrix (Pope and Mays 2007). The matrix, presented here, contains one overarching theme and three subthemes.

Results

The overarching theme, *It's hard for us*; *it's hard for them*, acknowledges the challenge participants addressed surrounding the identification and disclosure of incidental genomic findings that involve children. Although this was a unified theme across all of the stakeholders, there were distinctions that separated the professional stakeholder groups (IRB chairs, clinicians, and researchers) from the lay public groups. These distinctions appeared to cluster as if they were in response to three questions: (1) What to disclose?, (2) Who gets the information?, and (3) What happens later? Accordingly, the results are presented using a point/counterpoint format, with the professionals representing one point and the lay public representing its counterpoint (Table 2).

Subtheme no. 1: what to disclose?

Professionals: "it's complicated"

Professionals were cautious as they spoke about evaluating and/or returning incidental or unsolicited genomic findings that involved children. One reason given by professionals for this caution was their perceived inability to provide answers. As one professional explained, "We don't have an exact answer and I think it's difficult when you don't because the patient wants an answer… to help them make an informed decision." Another professional reiterated, saying "There's a huge stress component for everybody because *we*

Table 2 Thematic matrix—one overarching theme and three subthemes

Overarching theme	It's hard for us; it's hard for them	
(Subthemes)	Professionals	Lay public
What to disclose?	It's complicated	Lay it on the table
Who gets the information?	Complex dynamics to consider	Parents as gatekeepers
What happens later?	Following the child—a logistical nightmare	It's on the parents

don't know." The various professionals spoke about the proliferation of genomic knowledge and the increasingly complex nature of interpreting information surrounding an unsolicited genomic finding. As one professional shared,

> "I have a child with some dysmorphism or some developmental delay and I'm just going to see what's out there… so it's not the notion of not only dealing with any incidental finding but it's also sitting down and thinking, okay, we've got three things here… which of them is incidental, which of them is causative. How sure am I… then how do I deal with the causative… then how do I deal with the incidental, how do I classify it… do I even know what kind of risk it poses… it just really opens you up to all kinds of findings that you couldn't possibly expect and you couldn't possibly help the family or patient anticipate."

Multiple issues were also raised with respect to children, including, but not limited to, current vs. future accuracy of genomic tests in predicting associated phenotypes, especially given reduced penetrance and variable expressivity. As one professional shared, "You can't really predict the phenotype… so this was very difficult for the family… it was difficult for everybody concerned." Other issues suggested variability in timing or onset of phenotypic expression, especially when the incidental genomic finding pointed to adult-onset disorders (e.g., Huntington disease) or heightened gene–environment contributions to disease risk or health.

While the prospect of interpretive ambiguities gave rise to some hesitancy among clinicians, researchers, and IRB chairs, the professionals appeared most comfortable when the incidental finding was assumed to be clear and actionable. As one professional stated, "When you have an incidental finding that is alarming and turns out to be the real deal and significant… you're mostly worried about the extra complexity of communicating accurately the significance to the parents." There was a general agreement on sharing life-threatening or treatable conditions, especially when the treatment needed was more immediate or timely. However, the farther out from childhood the condition was expected to surface, and/or the absence of any available treatment gave this group some pause. As one professional cautioned, "It's children… at the beginning of their lives," while other reflected that, "You have to pick and choose which things to discuss with the family" and "It can impose a source of anxiety for which there is no treatment… that is, no treatment for the anxiety."

Yet, despite professionals' hesitation in predicting how they would react to incidental genomic findings in children, specific management suggestions did emerge in the professionals responses, including the need to share: (1) what one is looking for (why *this* test in this individual); (2) what one might also find out (using this test) directly about the individual, or indirectly about a family member, that could have immediate or future clinical significance; and (3) what you have is no way of predicting beforehand. One professional shared a suggestion about how to explain to parents just how complicated things can get, by saying, "We may come out of the situation after testing with results that we don't know exactly how to interpret…"

Lay public: "lay it on the table"

Members of the lay public were matter of fact, stating that they were willing to embrace any ambiguity surrounding incidental genomic findings, as they would to any other ambiguity, as a part of life. The bottom line seemed to be that the lay public participants expected practitioners/researchers to share information with them, whether the information was about themselves or their children, because having information is helpful. When asked about incidental findings involving children, one individual stated, "Just lay it on the table," while another suggested, "Give us the information… we'll do what we can."

The public members wanted to be informed upfront, *before* testing, of the possibility of incidental findings. Participants suggested that, in general, they would be more suspicious that something significant had already been found, if the choice to be informed about an incidental finding was only offered to the parent *after* the discovery was made. As one individual explained, "If you are asked after the fact… you've got them thinking, okay, they found something." As one individual said, "It's always good to know ahead of time… so you can think… Do I wanta do this?… Do I wanta take that risk?" Another member of the public summarized by saying, "For them to want to do all these tests, there's gotta be something making them think that maybe it's this, or maybe it's that, so just tell us." The emphasis on obtaining information was to be prepared. Lay public participants also made specific management suggestions with regard to managing incidental findings. Of note was that the only variation noted when the individual involved was a child was an almost uniform deferral to the child's parents. For example, participants suggested that the practitioner/researcher tells the parents what was being looked for and what information might also be found and that there

was a possibility of finding other types of unexpected or unsolicited information. These suggestions mirrored the management strategies proposed by the professionals. The deferral to parents as gatekeepers is described further in the next point/counterpoint regarding who gets the information.

Subtheme no. 2: who gets the information?

Professionals: "complex dynamics to consider"

When children are involved, most professionals stated that the dynamics were more complex, especially in terms of preexisting family dynamics, communication patterns, and parental gatekeeper issues. The professionals also shared concerns that children might be variably impacted, labeled, treated differently by parents, or even robbed of a normal childhood. As one professional asked, "Will this incidental finding have a beneficial or deleterious impact on the healthcare management of that child over their lifetime?" and "How will it impact their relationship with their peers… their parents, and their own self-image?" Another professional stated, "You worry that something you say will change the way that family thinks about that child, or that parent thinks about the child… that you will make the child abnormal in some way because they had this thing that the parents really don't understand and that you haven't explained very well because you don't understand." Another professional reiterated, "When it's a child… not only are you causing distress to the parent, but you are potentially altering the type of nurturing or upbringing that child is getting and, maybe, even altering the choices the parent offers or encourages when the child is growing up."

Some professionals specifically identified variables to consider, including inter-parent and/or family dynamics, which could complicate and possibly backfire with a one-size-fits-all approach. For example, one professional spoke about complex decision making surrounding incidental genomic findings in children, saying "I mean… if it's a young kid, there's too many variables there to consider," while another spelled out some of the complexity,

> "One is that the children themselves are not making the decision with regard to whether they get the information, so you have to think, you have to involve parents or guardians… there's that whole one parent, two parents… are they acting in the best interest of the child… the age of the child is important, so is this something that you should share with the child, something that you should share only with the parents…"

Most professionals acknowledged that parents were the logical gatekeepers for information surrounding the discovery of pediatric incidental findings, stating that parents have their children's best interests in mind when making decisions. As one professional stated, "You need to tell the parents and let them handle it," while another pointed out, "Children are unable to make informed decisions for themselves… so you'd want to make sure that they had an available parent or guardian who had the child's best interest at heart." However, cautionary reference was also made with respect to situations in which a parent(s)' judgment could be questioned. For example, one professional said, "I mean you tend to defer to the parent's judgment, but that can sometimes be a frightening concept."

While the view of parents as gatekeepers was agreed upon, the importance of including the perspective of the child was also alluded to. For example, one professional asked,

> The parents say no, we don't want to know about it… does the child have the right to make that decision when they come of age… because unlike a drug study, where the drugs are done and they're gone, genetic findings exist for the rest of that (child's) life.

One professional asked, "What type of information should be the child's to decide?," while another commented on the lack of evidence to guide them, "Nobody really knows what the impact is of telling children these sorts of things… we all just assume that either it doesn't make any difference, or it's bad, it's going to worry the child… but there's no evidence." Some professionals suggested waiting until the child was 18 or 21, for example, saying "We may want to wait for that test to be done when the child becomes an adult and able to make the decision whether they want to have testing that impact their insurance… or their life."

Another set of cautionary qualifiers emerged around parents' literacy and ability to understand the complexity of genomic information. For example, one professional said, "You may tell somebody that they have a genomic finding that may be associated with a potential for disease onset, but it may be very hard to explain to somebody who is a lay person how that risk really impacts their life," while another said, "We tell them that, but whether or not they hear, it is a whole other story." One professional pointed out, "Families have a hard time making decisions… (asking) What do (we) do with this?… What does this mean for me and my children?," while another reflected, "You do a test for all the good reasons, but it may not necessarily make everybody's life easier."

A final issue under this subtheme was the need for consideration of reproductive utility. Professionals noted that an incidental genomic finding in a child might have implications for parents, as well as for other family members as they consider future pregnancies. While being not necessarily significant for the child, the finding might be important for that child's future offspring's health and therefore would need to be shared before the child became sexually active. As one professional commented, "Well, there's the reproductive risks for them when they grow up… hopefully by the time they're

older, we'll have a little more information about that particular variation and what it means for them."

Lay public: "parents as gatekeepers"

The public members were not only ready to embrace the ambiguities that came with incidental findings when the individual involved is a child, but they were also clear in their identification that parents were children's primary gatekeepers. The lay public participants were unequivocal in their stated beliefs that parents are the rightful gatekeepers of children's genomic information, deciding if, when, and how information should be shared with their children. As one individual shared, "You would have to get approval from the parents to be able to share that information." The public members commented that parents are expected by the society to be responsible for their child's well-being and for their child's medical decisions. They emphasized that parents know their own children best and should therefore be deferred to as the experts, in terms of their children. One individual stated clearly, "The difference is… it's *my* child." As another shared,

> "I think the parents should know, because they're the ones that're looking after their child and they're the ones that are gonna see their progress… only a parent knows when your child is mature enough to accept this kind of information."

Notably, some participants suggested that parents may need help in explaining things to their children and should be able to return to either the research team or their child's health-care provider for assistance. Participants described working in partnership with health-care providers and researchers, stating, for example, that parents may (1) need to be reminded to talk to their children, (2) need help explaining, or (3) want to defer the discloser role to the clinical or research professional. They also shared that children be given the opportunity to ask questions that children might not want to ask their parent. For example, one participant said, "I'd want my child to be able to talk to the doctor and, you know, if they had extra questions or things that I couldn't answer then I'd want them to feel comfortable talking to the physician."

Subtheme no. 3: what happens later?

Professionals: "a logistical nightmare"

Professionals emphasized the challenge of situating incidental genomic findings amid burgeoning information, not only about genomic variants but also about disease causation, risk, and gene–environment interaction. As one shared, "I think our technology is advancing so rapidly that we don't understand what we're actually seeing sometimes." However, the greater concerns across professionals surrounded the almost overwhelming logistics of following a child over her/his lifetime. One professional equated it to opening, "a whole new can of worms."

While professionals agreed that parents serve as surrogate decision makers until children can make their own decision, they acknowledged a need to reassess children's developing autonomy as they move toward adolescence and young adulthood. Many professionals felt that information about incidental genomic findings in a child should be given to the parent(s) initially, thus shifting the "burden" of disclosure and follow-up to the child's parents. Although participants also suggested the gradual inclusion of children as they "matured cognitively," there was no uniform response regarding the age at which this gradual inclusion would occur or how cognitive maturity would be determined. There was, however, agreement across professionals that the logistics of keeping up with advancing genomic knowledge and children's developing autonomy was overwhelming, as each of these factors was seen as a "moving target." Some of the specific recommendations provided by professional stakeholders included (1) begin to engage the child at age 7 (the age of assent), (2) increase engagement when the child is 14, and (3) recontact the child/family when the child turns 18. However, there was no overall consensus.

Lay public: "it's on the parents"

The public assigned the responsibility of keeping track of information about a child to the child's parents. As one lay participant said, "I think you have to trust (the parent)… unless there's (you know) a history of obvious questionable judgment or lack of proper care or something." The public pointed out that the parents should take on most of the responsibility for not only making sure the information is recorded but also for making sure that information follows the child. As one stakeholder stated, "Put more responsibility on patient for keeping track of this information… it's fine if it's in your medical record, but it's your responsibility to make sure that it gets transferred." While another reiterated, "I think that'd actually be out of the doctors hands… the doctor could be (like) look, I tried y'know… (but) there's nothing he could really do about that… it's on the parents."

While the lay public generally viewed parents as the primary gatekeepers of information, especially adult-onset incidental genomic findings, they placed a time limit on the gatekeeping. They suggested that professionals serve as the backup but give parents a time deadline for disclosure upfront, saying, for example, "Hey you guys need to tell 'em at this point in time… or I will." Participants shared that, once a child is 18 years old, the professional becomes obligated to disclose information that may affect the child's future health or reproductive decisions. As one lay public participant shared,

> "I don't think that there's any way that a professional can be assured that parents will talk with their children… other than, you know, cross their fingers and hope that the parent chooses to be responsible. Perhaps the information is something that professionals could hold onto and, at the age of 18, … send that child a letter… or call the parent and say have you discussed this with your child. If it's something important that they (children) do need to know about, you're obligated as health-care providers to, you know, let the children know what they're at risk for."

Another possible backup or safeguard alluded to was a "permanent" health-care record that would follow the child into adulthood, especially when parents forget, die, or are unavailable, as may occur with foster placement or adoption. As one individual pointed out, "I think, it should probably still be stored in a file of some sort because, for example, I don't even know if I had chicken pox when I was little." The public viewed the medical record a reliable place to store information about incidental findings. Placing the information in a medical record seemed especially important for children because the information may not affect the child or be relevant for a long time.

Some individuals added that, while parents could be viewed as the primary gatekeepers and the ones with the ultimate responsibility with respect to disclosure of information surrounding incidental genomic findings, providers and researchers also bore some responsibility to monitor parents. For example, one explained, "It's something you could hold onto… and at the age of 18, send that child a letter or call the parent and say have you discussed this with your child?" One individual even equated a lack of responsible gatekeeping to child neglect, saying, "Parents are obligated to let their children know about genetic information and to use that information to take care of their children… if parents do not use the information responsibly, it should be grounds for neglect… parents should be held accountable."

Discussion

The purpose of the current study was to capture any unique issues and challenges surrounding the discovery and disclosure of incidental genomic findings when the individual is a child. Such insight is not only important to the development of future policies and best practice but also provides empirical evidence to support the growing number of position and reflective papers (e.g., Hens et al. 2011; Wilfond and Carpenter 2008; Wilfond and Diekema 2012). In this section, we discuss our findings, suggest two theoretical lenses through which to view them as we proceed forward, and highlight the major limitations we encountered.

Our primary finding was that public and professional perspectives diverged on the topic of disclosing incidental genomic findings that involve children. However, rather than considering this divergence as a barrier to dialogue, we propose that, as presented in the thematic matrix (Table 2), the questions and professional and lay public responses can serve as a topic guide for future inter-stakeholder dialogue focused on increasing understanding or shared meanings between these groups. Such an effort could raise awareness of taken-for-granted assumptions and encourage future collaboration that, in turn, is needed to inform the best practice regarding the disclosure of incidental genomic findings that involve children (Zorn et al. 2012).

The perceived issues and concerns surrounding disclosure of incidental findings involving children included (1) parents serving as gatekeepers, (2) the limitations/challenges of *substituted* autonomy and parental *proxy* consent, (3) ongoing assessment of children's developing autonomy, (4) preexisting family dynamics and communication patterns, and (5) logistics involved in following a child over time, including the potential need to provide a parental backup system and/or revisit the consent process at 14 and/or 18 years of age. These issues are not new. Others have written about the particular challenge for pediatric providers and researchers that accompany substituted autonomy and proxy consent (e.g., Wilfond and Diekema 2012). However, of particular note was the degree of responsibility that lay public participants were prepared to grant parents in terms of decision making around the receipt (*parents as gatekeepers*) and ongoing management (*it's on the parents*) of incidental genomic findings regarding their children. While professionals agreed that parents were the primary gatekeepers of information, there was hesitation about selecting what information to share with parents in the first place. This hesitation among professionals may reflect reluctance toward the wider trend toward adoption of transparency and shared decision-making models (e.g., Elwyn et al. 2012). It may also reflect professionals' recognition that the parental gatekeeper role is time limited and that a disclosure decision may best be made with the child as she/he develops. The recommendation to disclose findings to both the child/adolescent and parent, especially in handling incidental findings of clear and proximate clinical importance, has already been made (Wilfond and Carpenter 2008).

While professional stakeholders paused to contemplate disclosure, pointing to the complexity of data, nuances and ambiguity in interpretation, and concern about the potential for harm, the lay public participants seemed to embrace complexity and ambiguity as concepts that are already a part of their everyday life. The bottom line in the lay public group was when it comes to information with respect to themselves (Daack-Hirsch 2012) or their children; they wanted the option to know about it. A similar finding has been reported in other publications engaging the lay public (Tercyak et al. 2011;

Townsend et al. 2012). Lay public participants also felt that parents were the best recipients and keepers of information pertaining to their children. However, they did suggest that parents may need support or assistance deciding what findings to disclose, as well as how to disclose them to children at different ages. This need for professional assistance in explaining genetic concepts, including disease causation/risk, to their children has also been reported in other studies (e.g., Gallo et al. 2005; Metcalfe et al. 2011). A separate backup system or permanent record of findings was also suggested by the lay public in case something happened to parents before they were able to share anything with their children. In some cases, the backup system could also serve to remind parents upfront that their role as gatekeepers was time limited.

The bottom line for the professional stakeholders was focused on deciding what, or even if, incidental genomic findings should be disclosed to parents in the first place, especially if there was uncertainty about the interpretation of the finding. Their concerns appear rooted in a sense of responsibility for burgeoning amounts of genomic information and the interpretive challenges that follow. There was also a do-no-harm undercurrent in professionals' perspectives on children's immediate and long-term psychosocial well-being. This perspective was interesting because there is continued evidence that children who receive information from genomic-based tests and screening, whether indicative of increased risk or not, do not experience significant changes in psychosocial well-being or functioning (Wade et al. 2010). Going forward, the apparent hesitation of professional stakeholders may increasingly be influenced by emerging trends in clinical practice toward transparency of medical records and care (Delbanco et al. 2010).

As we considered our next steps, we reviewed the literature for alternative theoretical lenses that might provide different approaches or mechanisms for future investigations and/or interpretations. Two theoretical lenses stood out: Guillemin and Gillam's "micro-ethics" lens (2004) and Petronio's "Communication Privacy Management Theory" (2002, 2008). According to Guillemin and Gillam, a micro-ethics lens is used to capture the moral complexities encountered in everyday medical practice, or *ethics in practice*, which are often overlooked when the focus is on formal procedures, regulations, and policies, or *procedural ethics*. Ethics in practice, as understood by Guillemin and Gillam, exist at the everyday and informal, rather than the procedural level. While ethics in practice are easily overlooked or lost in the routine of everyday practice, professional stakeholders in our study acknowledge that there is nothing routine about incidental genomic findings involving children. Instead, professional stakeholders shared the view that ethical dilemmas surrounding incidental findings in children were both visible and worrisome. For Guillemin and Gillam, such an intersection would suggest that the disclosure of incidental genomic findings is what they term an *ethically important moment* calling for thoughtful reflection and response.

Another theoretical lens that might provide guidance for such an ethical importance is Petronio's Communication Privacy Management Theory (CPT), which focuses on hypothetical maps in understanding the different ways people navigate privacy as well as establish and reshape privacy boundaries (2008). Petronio's CPT outlines the foundational criteria that affect personal privacy rules, highlighting that ownership and control of private information do not always coexist. Most relevant to our findings, and to the larger shift toward person-centered care and shared decision making, is this theory's guidance in negotiating mutual privacy rules between different types of stakeholders (e.g., professionals and lay public). What may also be helpful in future research and policy development is how this theory provides a framework for understanding what they refer to as boundary turbulence, created when coowners of information are unable to negotiate and/or follow mutually held privacy rules.

One of the limitations of this study was that children were not the primary focus of the study, and accordingly, participants were not specifically recruited because of their "parent" status. Instead, the larger study focus was on recruiting a broad cross-section of stakeholders, with only one group purposively recruited because they were parents of children who had undergone genetic testing. Despite this, what was astonishing was that across all of the lay public groups, participants identified parents as gatekeepers. While there were follow-up probes about the various responsibilities and challenging logistics, participants offered very few specific suggestions beyond a persistent reiteration that "something" would need to be worked out, and parents might need a backup system. Additional studies are needed to explore what could work for both parents and professional stakeholders in both clinical and research settings. Our analysis does not offer a solution but, instead, emphasizes the distinction between the two groups as a point for further research and interactive problem-solving conversations.

Another limitation was that while we anticipated the need to orient the lay public participants to the definition of incidental findings, we assumed that professionals shared our understanding of incidental findings. Instead, we found that some of the professional stakeholders held varying ideas about what constituted an incidental finding in a genetic/genomic study, which we have reported in more detail in another manuscript (Downing et al. 2013). In future studies and/or interactive problem-solving conversations, we would recommend that all participants agree on the definition of what constitutes an incidental findings first as the definition will be specific to the context.

Conclusion

This study is an early effort to engage multiple stakeholders in identifying issues and concerns with respect to incidental genomic findings involving children. All participants agreed that disclosure of genomic information about children is complicated and that the issues are bidirectional (It's hard for us; it's hard for them). We also noted that while the questions that need answers are clear and agreed upon, professional stakeholders and lay public participants more often have different perspectives. The differences, as well as the similarities or overlaps, in their answers, provide insight into the issues and concerns needing future dialogue. This dialogue, in turn, is needed not only to build understanding and trust between professional stakeholders and lay public participants on the management of incidental genomic findings in children but also to embrace the continued engagement of lay public participants. Such dialogue could also provide further empirical evidence to inform policies and best practice regarding disclosure of incidental genomic findings involving children that reflects the IOM's call for new approaches that focus on the patient or person's priorities and needs, rather than simply on how the larger research or clinical setting define its own needs, priorities, and outcomes.

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