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Exome sequencing disclosures in pediatric cancer care: Patterns of communication among oncologists, genetic counselors, and parents

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ABSTRACT

Objective: To examine communication patterns and behaviors during disclosure of exome sequencing (ES) results to parents of pediatric cancer patients, and describe common themes in parental communication.
Methods: Using mixed methods, we analyzed transcripts of sessions where parents of pediatric cancer patients received ES results from an oncologist and genetic counselor. Seventy-six transcripts were analyzed for frequency of clinician information-giving, partnering-supportive talk, and active parent participation. A subset of 40 transcripts were analyzed using thematic content analysis.
Results: Disclosures consisted mostly of clinician talk (84% of total talk), which was focused on providing information (62% of clinicians' utterances) with occasional partnering-supportive talk (7% of clinicians' utterances). Most parents assumed a passive, listening role (16% of total talk). Themes in parental communication included expressing relief and the significance of an answer, concern about sharing results and responsibility for inheritance, and seeking clarification of health implications of results.
Conclusion: Our finding of low levels of active parent participation during ES disclosures highlights the need to improve patient/parent engagement and understanding in a genetic setting.
Practice implications: Clinician communication strategies that could encourage parent participation and understanding include checking for parent understanding, partnership-building, and tailoring ES discussions to address parent concerns and preferences.

1. Introduction

Exome sequencing (ES) and other clinical next-generation sequencing tests have the potential to impact childhood cancer care through discovery of somatic mutations or germline cancer susceptibility mutations, in addition to clinically significant incidental findings unrelated to the test indication. [1,2] While studies have examined the perspectives of pediatric oncologists, parents and patients regarding ES through interviews and surveys [3,4], understanding the communication of ES results is still limited. A number of challenges when conveying genomic information can complicate communication. Most variants identified by either tumor or germline ES are not likely to be significant with respect to the child's current treatment [5]. Further, reports often include recessive and pharmacogenetic variants unrelated to the diagnosis but with implications for future health-related decisions (including family) as well as variants of uncertain significance (VUS).

In cancer consultations, patients and parents are often involved in asking questions, expressing concerns, and stating preferences

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as they make decisions about treatment and follow-up care. Clinicians, in turn, provide information about disease and treatment, attempt to assuage emotional distress, and (ideally) facilitate patient involvement in the consultation [6]. In genetic consultations, clinician talk can dominate [7,8] as foreign genetic concepts, and their clinical and psychosocial implications for patients and families, must be conveyed. Information giving must be balanced with genetic counseling and psychosocial support. Best communication practices for ES disclosure sessions in cancer care could help clinicians navigate these complicated encounters, but a more robust evidence base is required for their development.

We present data on the communication of germline ES results from a clinical trial of tumor and germline ES in newly diagnosed pediatric solid tumor patients at a large medical center [1]. We describe communication patterns within ES disclosure sessions that included the patient's primary oncologist, a genetic counselor (GC), and the parent(s). The study explored the distribution of talk as well as type of communication, including clinician information-giving, and patient-centered communication such as, partnership-building (e.g., facilitating or accommodating parent participation) and supportive talk (offering encouragement, empathy) [9–12], and degree of active parent participation (e.g., asking questions, expressing concerns) [13,14]. We also explored qualitative themes that emerged from the parents' communication during the disclosure sessions.

2. Methods

2.1. The BASIC3 study

The Baylor College of Medicine Advancing Sequencing in Childhood Cancer Care (BASIC3) study, funded by the US National Human Genome Research Institute and National Cancer Institute, is a Clinical Sequencing Exploratory Research program project [15] that examines the implementation and clinical utility of tumor and germline ES in the care of childhood cancer patients [1].

Study enrollment began in August 2012 and was completed in June 2016 with 287 patients and their parent(s)/legal guardian(s) enrolled (enrollment rate of 71%). Newly diagnosed brain or solid tumor patients, less than 18 years of age who were English or Spanish speaking and who had undergone surgery and were receiving care at the institution were eligible for the study. A total of 17 pediatric oncologists were also enrolled. The study was approved by the institutional review board (IRB) of Baylor College of Medicine (BCM) and Texas Children's Hospital.

During informed consent, members of the study team provided parents with detailed information on study events, the types of tumor and germline findings reported, findings for which parents had the option to receive or not receive, and the risks and benefits of participation [16]. Once enrolled, patient blood and tumor (if available) samples were submitted for clinical ES and reported in the child's medical record. Parental samples were collected when possible to aid in interpretation of germline findings. Oncologists attended two educational sessions, the first an orientation to both ES and the BASIC3 study design and the second focusing on the organization and content of ES reports. The study principal investigators (a pediatric oncologist and a medical geneticist) and study GCs reviewed ES results with the patient's primary oncologist prior to disclosure.

The ES disclosure sessions were audio-recorded and typically occurred in conjunction with a scheduled oncology visit. The oncologist and GC (referred to collectively as clinicians) were both present with the parent(s) or legal guardian, other family members or friends, and sometimes the patient. Although the oncologists were study participants, the GCs were study staff and served as resources to the oncologists, particularly with respect to discussing findings related to carrier status and VUS as some oncologists deferred to the GC's expertise for these results. The discussion of results was structured according to the categories in the clinical germline and tumor ES reports [1,17] (Table 1). Here, we specifically examined communication of germline results because (a) not all patients had a tumor report and (b) discussion of tumor reports tended to be much briefer. Germline results were reported by category as listed in Table 1, clinically significant categories are noted with asterisk. Paper copies of the ES reports and a genetic counseling letter were provided to families at the disclosure sessions.

2.2. Sample and data analyses

We conducted a quantitative analysis of 76 ES disclosure sessions completed between August 2012 and May 2015 to examine patterns in communication among clinicians and parents and a qualitative analysis of a subset of these same 76 disclosures to explore themes in parental communication. The 76 disclosures involved 11 oncologists. The remaining 6 enrolled oncologists either had fewer than four disclosures (a minimum threshold to limit variability) or joined the study after 2015. The number of disclosures analyzed per oncologist ranged from 4 to 13 (mean = 6.9, median = 4). Once it became clear that the preponderance of findings would not be significant with respect to the child's clinical care, the number of disclosures per oncologist was capped at 4 unless subsequent disclosure sessions had a significant finding. If so, it was added to the sample for analysis. The audio-recordings were fully transcribed and marked to designate germline from tumor discussions.

From these 76 sessions, the 16 disclosure sessions with a molecular diagnostic or incidental germline finding were selected for qualitative analysis along with 20 disclosure sessions without significant germline findings randomly selected from the remaining sample. Four additional disclosures with a molecular diagnostic germline finding that occurred after May 2015 were added to the qualitative sample to ensure thematic saturation, that is no new codes or themes emerged, had been reached across the types of results received [18], bringing the total qualitative analysis to 40 sessions and the total sum of disclosures analyzed to eighty.

2.2.1. Quantitative analysis

Clinicians' and parents' communication behaviors were coded using the Physician-Information Giving and Active Patient Participation Coding Systems, validated in previous studies [19,20]. Using the utterance (oral analogue of a simple sentence)

Table 1
Categories of germline variants.

Variant Category	Definition
Category 1*	Deleterious mutations in disease genes related to clinical phenotype
Category 2	Variants of unknown clinical significance in disease genes related to clinical phenotype
Category 3*	Medically actionable deleterious mutations in disease genes unrelated to clinical phenotype (secondary findings)
Category 4	Carrier status for recessive Mendelian disorders
Category 5	Pharmacogenetic profile variant alleles
Category 6*	Mitochondrial mutations

* Categories considered to be clinically significant.

[21]as the unit of analysis, we coded three types of communication behaviors. Clinician *information-giving* related to a description or explanation of genomic findings or other health-related issues. Clinician *partnering-supportive talk* which consisted of behaviors supporting parent involvement and included partnership-building (e.g., "What stands out about the information?"; "What concerns would you like to discuss?") and supportive talk (e.g.," You all have shown strength during this time," "Yes, it must be very difficult for the family") [20,22]. Parents' *active participation* behaviors in the disclosure visits included asking questions, assertive comments (e.g., making a request, stating preferences), and expressions of concern (e.g., worry, fear).

Oncologist and GC behaviors were summed to create a composite clinician communication score for information giving and partnering supportive talk for each disclosure session. As in other cancer care settings where patients were accompanied [23], one score also was computed representing the parents' collective communication. Three coders participated in three 2-hour training sessions. Once coders achieved consistency, transcripts were independently coded; 15% of the disclosures were coded by more than one coder with agreement assessed by intraclass correlation coefficients (ICC), which ranged from 0.68 to 0.89 indicating acceptable reliability.

To control for factors potentially associated with variability in clinicians' communication as well as parents' active participation, data were analyzed by generalized linear models using Poisson distribution with a log-link function. An exchangeable working correlation structure was specified in the models and oncologist was included in the models as a random effect. Predictor variables included presence of a significant genetic finding, oncologists' experience presenting the report (first two vs. third or more), number of adults in attendance (1 vs 2 or more), and primary parent's self-reported ethnicity and race (Hispanic/Latino (all races); African-American/Black, non-Hispanic White, and other). Because clinicians' partnering-supportive talk has been associated with greater patient involvement in consultations [20], it was also included as a predictor of active parent participation. All analyses were conducted using SAS 9.4 (SAS Institute Inc., Cary, NC, USA).

2.2.2. Qualitative analysis

Four members of the research team identified themes from disclosure transcripts following standard methods for team-based qualitative analysis and consensus coding [24,25]. Analysis focused on exploring themes of topics raised by parents (e.g., questions, concerns, expressions of preferences) related to disclosure of their child's germline ES results. Analysis did not focus on themes of provider utterances. A coding scheme was developed deductively based on key research questions and further refined by an inductive approach to identify recurrent themes [26]. Transcripts, codes, and memos were entered and managed using QSR international's NVivo 11 software. Data were analyzed for common themes across all disclosures as well as for distinctive themes within disclosures with significant versus non-significant findings.

3. Results

Table 2 provides demographic characteristics of the primary parent (self-defined by each family at study entry) included in the sample, which are consistent with the highly diverse nature of the TCH patient population [16] and race/ethnicity of the study oncologists. Age of oncologists was not collected. Across the BASIC3 study (n = 287), approximately 13% of subjects had a pathogenic or likely pathogenic variant in either a diagnostic or medically actionable incidental finding, which was considered clinically significant [1]. In contrast, there was a median of three VUS and two carrier status findings per germline report making them the most common findings returned to participants.

3.1. Distribution of talk and communication patterns

Clinicians accounted for most of the talk in the disclosure sessions (84%). Clinician communication focused mostly on providing information (62% of clinicians' talk) with occasional partnering-supportive talk (7% of clinicians' talk). Use of partnering-supportive talk increased significantly after the clinicians had conducted at least two disclosure sessions (OR = 2.9, P < .001). With a few exceptions, most parents assumed a relatively passive, listening role in the disclosure sessions, accounting for just 16% of the total talk. Parents averaged 7.8 (range 0–54) active participation utterances which represented 19% of their total utterances. Of the active participation utterances, 74% were in the form of questions, followed by acts of assertiveness (20%). Only 6% of the parents' utterances were coded as expressing concerns.

A parent was accompanied by another adult (usually another family member) in the majority (78%) of the disclosures. When more than one adult was present, clinicians provided more

Table 2
Parent, oncologist and disclosure visit characteristics.

Characteristic	N / Mean	% / Standard Deviation
Primary Parents (n = 80)		
Age	36.5	7.3
Race/Ethnicity		
- Non-Hispanic White	37	46%
- Black	12	15%
- Hispanic	22	28%
- Other	6	7%
- Unreported	3	4%
Study Oncologists (n = 11)		
Race/Ethnicity		
- Non-Hispanic White	5	46%
- Black	1	9%
- Hispanic	2	18%
- Other	3	27%
Disclosure Visits (n = 80)		
Disclosures with >1 parent present	62	78%
Disclosures with significant germline finding(s)	27	34%

information (P = .023) and engaged in more partnering supportive talk (P = .021) (Table 3), and greater active parent communication was observed (P = .047) (Table 4). Clinicians provided more information when there was a significant clinical finding (P=<.001) (Table 3). However, parents were no more likely to ask a question or express concerns when there was a clinically significant finding than when such a finding was absent (Table 4).

Active parent participation did not differ by race/ethnicity in germline ES disclosures (Table 4). However, clinicians engaged in less partnering-supportive talk when the primary parent was non-Hispanic Black/African American than when talking with non-Hispanic white parents (germline P = .013) (Table 3). While worth noting, given the small sample size (n = 12 non-Hispanic Black/African Americans) our sample is insufficient to make generalizations.

3.2. Themes in parental communication

Given that the amount of active participation by parents was limited and did not vary by result type, we performed a more in-depth analysis into the content of parental communication to identify significant themes.

3.2.1. Expression of relief and seeking reassurance

Parents expressed relief about study results, whether a significant finding was returned or not. As illustrated by two parents receiving the diagnosis of a cancer predisposition syndrome in their child, results led to relief at finding a cause for the cancer and having direction for next steps in the care of their child and other family members. *"This is still sort of—to me it's good news. You know—good news because at least now I—we at least know who to test" (*906824*).* Some parents expressed that this was better news than they were anticipating, highlighting the expectation of receiving multiple clinically significant results. *"I'm actually relieved. I thought there'd be lists of things" (*1357902*).* Even parents whose utterances suggested surprise or shock transitioned to affirming the value of the information:

> Clinician: What are some of the main questions that you have?
> Mother: I can't really even think about that [genetic result] right now. That just kind of caught me off guard . . . .But it's something I've needed to know, and now I don't have to live in fear regarding it anymore. (176688, mother of child identified with a cancer predisposition syndrome previously suspected in family)

Table 4
Multivariate analyses of predictors of parent active participation.

Covariates	Utterance Ratio (95% CI)	p-value
Number of Parents in Attendance		
>1 parent	1.82 (1.01 - 3.29)	0.0468
1 parent	1	
Number of Reports Oncologist Disclosed		
≥3 reports	1.05 (0.63 - 1.76)	0.8486
1-2 reports	1	
Race/Ethnicity		
Black	1.23 (0.55 - 2.73)	0.6145
Hispanic	0.81 (0.35 - 1.87)	0.6279
Others	1.15 (0.67 - 1.99)	0.6118
Non-Hispanic White	1	
Genetic Significant Finding		
Yes	1.42 (0.86 - 2.36)	0.1736
No	1	
Patient-Centered Communication		
Yes, >median	1.81 (0.97 - 3.39)	0.0613
No, ≤median	1	

Parents also expressed relief when the information from genetic testing did not suggest a hereditary cause. However, some parents still wanted information about non-hereditary causes for their child's cancer. They also reflected on their desire for reassurance of the future health of their child and the inability to achieve this.

> Mother: I think the answer that we want is the answer that no one can provide. It's going to go away, it's going to stay away and it's never going to come back. And no one here can provide us with that . . . (55606, mother of child without a diagnostic germline finding)

3.2.2. Expression of concerns and sense of responsibility

arents given significant ES findings also expressed concerns about sharing results with family members. Concerns increased when a relative was known to be particularly anxious, had limited financial resources, or such that they might be burdened by the information.

> Mother: I'm trying to think about how to phrase this to all of the younger kids because I know my sisters are going to freak out, and my little cousin doesn't comprehend stuff when you use big words. (176688, mother of a child with an inherited cancer predisposition syndrome)

Table 3
Multivariate analyses of predictors of clinicians' communication patterns.

	Information-Giving		Partnering-supportive talk	
Covariates	Utterance Ratio (95% CI)	p-value	Utterance Ratio (95% CI)	p-value
Number of Parents in Attendance				
>1 parent	1.3 (1.04 -1.64)	0.0233	1.53 (1.07 - 2.21)	0.0211
1 parent	1		1	
Number of Reports Oncologist Disclosed				
≥3 reports	1.08 (0.88 - 1.33)	0.4509	2.9 (1.71 - 4.95)	<.0001
1-2 reports	1		1	
Race/Ethnicity				
Black	1.08 (0.72 - 1.61)	0.7019	0.56 (0.35 - 0.89)	0.0132
Hispanic	0.84 (0.67 - 1.06)	0.1372	1 (0.64 - 1.57)	0.9832
Others	0.94 (0.68 - 1.29)	0.6893	1.34 (1.05 - 1.73)	0.021
Non-Hispanic White	1		1	
Genetic Significant Finding				
Yes	1.44 (1.21 - 1.71)	<.0001	1.44 (1.08 - 1.93)	0.0132
No	1		1	

If significant findings were not returned, parents inquired whether there would still be a contribution for future cancer families from their participation in this research.

Mother: I think my main concern is I realize that even if the information doesn't help her, I want to make sure that the information will make an impact somewhere down the road. (39033, mother of a child without a significant finding)

A unique component of returning ES results is the potential to deliver results relevant to both the child and parent with one test. In these scenarios, families frequently made comments related to inheritance and their perceived responsibility while handling the news with humor or playful sarcasm toward self or partner:

Clinician: And that gene, like I said, has a fancy name, [GeneX]. And, it's a change that came, from—

Mother: One of us.

Clinician: One of you—let me see—came from Dad.
Mother: Good job. (laughter) (1077445, parents discussing inheritance of carrier status)

3.2.3. Seeking clarification of concepts and implications of results

In a number of comments, parents wanted to clarify and understand the information provided by the clinicians. As noted above, carrier status was the most frequent germline ES finding (approximately 90% of BASIC3 parents elected to receive this finding) [16] and prompted parental questions and statements requiring clarification, which is consistent with the quantitative findings indicating that 74% of the parents' active participation was accounted for by asking questions. While some parents were familiar with the general concept, others struggled to understand nuances such as recessive and dominant. "*Right. The recessive genes make the dominant? Correct? Isn't that right?" (*314485*).* Parents sought confirmation that this was something that their child would not develop. *"Okay, so just because you're a carrier don't mean you're going to have it? 'Cause I know I've never had it."(*1067068*).* Although parents sometimes inquired about steps they could take for their future pregnancies, the focus tended to be on the implications of this information for the child in the future.

Father: We're going to have this conversation with him?

Mother: And we're going [to] send up his potential wife to see—? (laughter)

Clinician: We could offer her this genetic test.
Mother: Make sure that my grandchildren are going to be okay? (314485, parents discussing carrier status finding)

Conversations about VUS in cancer genes could involve back and forth between parent and clinician about the concept that a VUS may have no significance or could potentially have clinical significance in the future.

Mother: Okay. But if she develops something like that, then we can go back and say, well – you know – she had this and this and they can say, 'Oh, okay.' That would then link the two.

Clinician: Over time, we may understand more about these genes as we do more tests like this. But right now, there's nothing to tell us that she has any of these susceptibilities or anything clinically that we would do to change our follow up with her.
Mother: Okay. So this is just an explanation; this isn't what she has . . . this is just stuff she could have. (421178, mother discussing VUS finding)

Some parents sought to attach VUS or carrier findings to known health conditions in themselves or their families.

Clinician: They can have things like skeletal problems, too—and hearing loss goes along with that condition.

Father: Scoliosis, too?

Clinician: Not as much—they can have some changes in their spine—not necessarily scoliosis.
Mother: Because I have that. (129921, parents discussing carrier status)

Even after dialogue addressing these misperceptions, parents could be persistent, raising these issues again later in the disclosure session, which were represented in some utterances coded in the quantitative analysis as assertive acts of participation.

4. Discussion and conclusion

4.1. Discussion

This study examined quantitative and qualitative communication characteristics of clinical interactions in which clinicians and parents of childhood cancer patients discussed ES germline reports. Because this is a novel context for ES, the findings have important implications for enhancing communication in this clinical setting.

Consistent with observations in genetic consultations [8,27,28], clinician communication in this context focused primarily on providing information. Pre-test counseling sessions include discussion about natural history, inheritance and medical management for the condition of interest. In genomic sequencing, post-test counseling can resemble this model of information-giving since the scope of conditions analyzed is too vast to review in detail at pre-test visits. Therefore it is of note, that the overall parent communication, and specifically active participation in this context was, on average, low when compared to patient/parent communication observed in other cancer [29], pediatric [30], and cancer genetic counseling consultations [7,31]. However, the reasons for this requires future research. For example, parents may be reluctant to ask questions, clarify any confusion, or feel overwhelmed given the amount and complexity of ES information [32,33]. Of note, active parent participation was greater when more than one parent or adult was present; thus, clinicians should encourage both parents (or an additional adult) to attend these sessions whenever possible.

Our results revealed good-faith efforts to check for parent understanding, a type of partnering-supportive talk. However, these were typically close-ended queries (e.g., "Does that make sense?" "Have any questions?") that often prompt only simple "yes" or "no" responses [34] which clinicians may erroneously interpret as comprehension [35]. Communication strategies such as 'ask-tell-ask'[37] and concern elicitation [36] are open-ended efforts to assess comprehension and identify concerns by providing reflective, explicit probes (e.g., "What do you find important about what we told you?" "What stands out to you about this information?") to better elicit parents' sense-making of the information. Parents intimidated by complexity may be more willing to speak up if clinicians normalize confusion (e.g., "many people find this confusing . . . "). In addition, recognizing common topics of concern expressed by the BASIC3 parents (concern about impact on their children and themselves, parental responsibility) can provide clinicians with guidance for initiating discussion with parents. These suggestions may be particularly helpful when addressing the preferences and communicative experiences of racially and ethnically diverse patient populations.

The devotion of proportionally more discussion of less direct clinically relevant findings (carrier results, VUS) undoubtedly reflects the fact that reports included findings in these categories. Further, the clinicians often followed the report order with VUS as the second category. An arguably more effective disclosure order would be findings related to the patient phenotype, other medically actionable findings, carrier findings, and finally VUS. Carrier status may be most relevant in families where parents are still in their reproductive years or older siblings are actively planning pregnancies. Of note, a multi-institutional pediatrics and genetics committee has recommended dividing up ES disclosure into multiple sessions to avoid information overload for parents and to distinguish different types of results [37]. However, the feasibility of multiple sessions for parents of seriously ill children undergoing complex treatment is unclear.

Despite the overall parental passivity, our thematic analysis identified topics that did engage parents. When receiving significant findings, parents explored its meaning to the health of their child and other family members and expressed relief with having answers about the cause of the cancer. For inherited variants, parents raised concerns about communicating findings to family members, asked questions about integrating this information into clinical care, and conveyed a sense of responsibility. In the absence of significant findings, parents often expressed relief, yet were left with unanswered questions regarding why the cancer occurred.

Parents also had questions when trying to understand what findings of carrier status and VUS mean in the context of their child's heath. Although clinicians attempted to correct misperceptions, this was challenging, especially when parents latched onto a clinical feature and asserted its relationship to their own health history, a phenomenon that has been observed in other genetic counseling situations [38,39].

This study has limitations. We did not determine the clinical impact of specific communication elements or assess parents' satisfaction with the disclosure session. Also, our study focused on parent-provider interactions only and did not analyze patient interactions given the diversity in age and participation levels of pediatric patients. Third, future studies should explore differences in oncologists and GCs comfort in discussing ES findings. Further, our study focused on a single center, which may limit the generalizability of the results and,while diverse, our sample was relatively small. Future research should examine communicative patterns and understandings in ES disclosures in multiple settings and include communication outcomes and psychosocial outcomes.

4.2. Conclusion

Our results provide important insight into an essential element of implementation of genomic medicine into pediatric cancer care: the return of ES results to patients/parents. Our findings highlight important areas for future investigations of communication of ES results, including best practices for enhancing patient/parent engagement, optimizing patient/parent understanding of ES results, and uncovering and addressing patient/parent concerns and preferences in this context. Although ES disclosure sessions involve the need for significant information-giving, parental involvement and understanding of ES results could potentially be improved by utilizing specific communication strategies that focus on the most relevant information and facilitate exploration of parental concerns.

4.3. Practical implications

To address challenges in eliciting parent involvement in sessions disclosing ES results, providers need to identify communication tools to encourage parental participation and foster understanding especially as genomic testing increases in complexity. First, clinicians can use a more open-ended approach to eliciting parental understanding and questions in order to identify misperceptions, topics of greatest interest to a particular family as well to identify families who may be overwhelmed by the information. Second, clinicians should have an awareness of topics that previous parents have noted to be of importance when receiving ES results. With this knowledge, providers can introduce topics for discussion with families who may not initially engage. Lastly, simplifying information that may be less relevant in a particular clinical context, such as carrier status and VUS, may help prevent confusion and encourage a clearer understanding of what is most relevant to the family. Future studies could explore the benefits of employing these approaches on the communication process in the context of genomic sequencing.

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Conflict of interest

Baylor College of Medicine (BCM) and Miraca Holdings Inc. have formed a joint venture with shared ownership and governance of the Baylor Genetics Laboratories which performs exome sequencing. Dr. Plon serves on the Scientific Advisory board of Baylor Miraca Genetic Laboratory. All other co-authors declare no conflict of interest.

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