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An Observational Study of Children's Involvement in Informed Consent for Exome Sequencing Research

Victoria A. Miller, PhD[a], **Allison Werner-Lin, PhD**[b], **Sarah A. Walser, BA**[c], **Sawona Biswas, MSc, MS, CGC**[d], and **Barbara A. Bernhardt, MS, CGC**[c]

[a]Division of Adolescent Medicine, Department of Pediatrics, The Children's Hospital of Philadelphia and the Perelman School of Medicine at the University of Pennsylvania

[b]School of Social Policy and Practice, University of Pennsylvania

[c]Division of Translational Medicine and Human Genetics, Perelman School of Medicine at the University of Pennsylvania

[d]Division of Human Genetics, Department of Pediatrics, The Children's Hospital of Philadelphia

Abstract

The goal of this study was to examine children's involvement in consent sessions for exome sequencing research and associations of involvement with provider and parent communication. Participants included 44 children (8–17 years) from five cohorts who were offered participation in an exome sequencing study. The consent sessions were audiotaped, transcribed, and coded. Providers attempted to facilitate the child's involvement in the majority (73%) of sessions, and most (75%) children also verbally participated. Provider facilitation was strongly associated with likelihood of child participation. These findings underscore that strategies such as asking for children's opinions and soliciting their questions show respect for children and may increase the likelihood that they are engaged and involved in decisions about research participation.

Keywords

assent; research; decision making; pediatrics; ethics

Both child assent and parental permission are required for children and adolescents to participate in research. The assent requirement can be waived if the research has the possibility of direct benefit to the child that is only available in the research context, or if the child is judged incapable of assent. The regulations are vague with respect to what constitutes valid assent, and prior research has focused primarily on children's understanding of research and the factors that influence their understanding (Dorn, Susman, & Fletcher, 1995; Fogas, Oesterheld, & Shader, 2001; Tait, Voepel-Lewis, & Malviya, 2003). Some research has focused on the relational aspects of assent, reflecting the view that collaborative decision making between parents and children is part of normative development (Liprie, 1993; White, 1996; Wills, Blechman, & McNamara, 1996). Prior

Correspondence to: Victoria Miller, The Children's Hospital of Philadelphia, 34th St. and Civic Center Blvd., CHOP Main, 11NW10, Philadelphia, PA 19104. millerv@email.chop.edu. Tel: 267-426-5259. Fax: 267-590-4708.

qualitative research has found that the majority of children and adolescents want parental input regarding research decisions or feel that the parent-child relationship plays an important role in these decisions (Broome & Richards, 2003; Geller, Tambor, Bernhardt, Fraser, & Wissow, 2003). A relational view of assent also recognizes that children and adolescents can be involved in decision making in multiple ways (Broome & Richards, 2003; Geller et al., 2003; Miller, Reynolds, & Nelson, 2008; Wilfond & Diekema, 2012). For example, children can express an opinion or ask questions about the decision to be made. Further, research personnel, providers, and parents can facilitate children's involvement by asking for their opinions about participation, providing information, or checking to make sure children understand aspects of research participation (Baylis, Downie, & Kenny, 1999; Joffe, 2003; McCabe, 1996; Weithorn, 1983). Being involved in these ways may help children to become better decision makers, enhance their self-efficacy (Liprie, 1993; White, 1996; Wills et al., 1996), and increase their satisfaction with the decision and adherence to research procedures.

Genomic sequencing research poses a unique set of challenges with respect to child assent. Families first may be asked to decide about research participation in which sequencing is performed to attempt to identify the cause of the child's medical condition. In addition, as a part of the research, they may need to decide on the categories or types of secondary findings they wish to receive, which may have implications for the child's future health status, as well as their developing identity and life plans (Sabatello & Appelbaum, 2015). When children and adolescents are not involved and/or their concerns are not addressed, the decision about exome sequencing may be dominated by the preferences and potential anxieties of the parents (Sabatello & Appelbaum, 2015). Furthermore, the results of genomic sequencing may have implications for other family members, pose the potential risk of discrimination, and have uncertain validity and clinical utility (Green et al., 2016; Wilfond & Diekema, 2012). A better understanding of how children and adolescents are involved in discussions about these complex issues and what concerns they have about sequencing will be a starting point for developing strategies to enhance the child assent process for genomic sequencing in both research and clinical settings.

The present study is a secondary analysis based on a larger study to bring genomic sequencing into a pediatric clinical setting and assess the impact of genetic testing on patients and families. A previously published analysis from this larger study utilized qualitative methods to examine adolescent engagement and decision making during 25 consent sessions of probands who were ages 12–19 years (Werner-Lin, Tomlinson, Miller, & Bernhardt, 2016). The analysis indicated that adolescents were mostly quiet during the consent session, and parents typically held roles as protectors and information-holders. In the present study we focused on children ages 8–17 years, quantified children's involvement, and assessed both provider and parent facilitation of children's involvement during the informed consent session. Given these different foci and methods, there is no overlap between the present study and what was examined and reported in the previously published qualitative study. The assessment of provider and parent communication during the informed consent process is important because children may need encouragement to be active participants in health-related decision making. However, such encouragement is understudied in the research context. The objectives of this study were to: 1) describe

children's involvement in discussions about enrolling in a research study of exome sequencing; 2) examine the association of children's involvement with provider and parent facilitation of involvement during the consent session; and 3) examine associations of children's involvement with age and duration of the informed consent session.

Methods

Recruitment and Participants

The study on which this analysis is based was conducted at the [name deleted for peer review] site of the Clinical Sequencing Exploratory Research Program funded by the National Human Genome Research Institute (Green et al., 2016). Participants were children and adolescents attending specialty clinics at The Children's Hospital of Philadelphia. A referring provider identified them because they had or were suspected to have non-syndromic hearing loss, cardiac arrhythmias or cardiomyopathy, nuclear encoded mitochondrial disease, platelet function disorder, or a neurodevelopmental disorder with a suspected underlying genetic cause.

Procedures

The study was approved by the IRB at [name deleted for peer review]. Study physicians (n = 7) from each of the specialty clinics, genetic counselors (n = 5), a nurse (n = 1), and/or a study coordinator (n = 1) met with families to describe the study. This could occur in the context of a new or follow-up clinical care visit or a separate research visit. Prior to starting the session, study personnel asked parents for written consent to audio-record the session for future analysis by the research team. Then, study personnel addressed the primary elements of informed consent for exome sequencing and study participation, including the procedures, risks and benefits, as well as the possibility of identifying results related to the child's primary condition ("primary" findings) and secondary findings (those unrelated to the child's primary condition). Study personnel informed the families that they would automatically receive immediately medically actionable secondary findings and were given the option of receiving secondary findings in three additional categories: childhood-onset non-immediately medically actionable conditions, adult-onset medically actionable conditions, and carrier status for recessive conditions. At the end of the informed consent session, parents provided written permission and consent for their children and for themselves to participate, and children who agreed provided written assent if they were capable of doing so. Assent was waived for children primarily presenting with neurodevelopmental disabilities or children who were medically considered to be incapable of providing assent by the referring clinician. Additional study procedures related to completion of questionnaires and the return of results to families are not relevant to the present analysis.

Measures

Demographics—Parents completed a demographics form that documented characteristics of the child, parent, and family, including child age, sex, and race, and parent marital status and highest educational attainment.

Observed Child Involvement—Study personnel downloaded digital recordings of informed consent sessions to secure servers and imported them into NVivo 10, a qualitative data management software program (*NVivo qualitative data analysis software*, Version 10, 2012). None of the study personnel who conducted the consent sessions were involved in the transcribing or coding process. A member of the research team listened to the audiofile and transcribed sections of the audiofile in which the child spoke or the parent or study personnel spoke to the child. The first author, in conjunction with team feedback, drafted an initial coding scheme, developed for this study, to capture aspects of child involvement, as well as provider and parent facilitation of child involvement. Two of the authors then coded several transcripts, and the team met again to discuss dissimilar codes and adjust the coding scheme. The same two authors then coded two transcripts independently and met to compare coding and resolved discrepancies. One investigator then coded the remainder of the transcripts. The final child, parent, and provider codes are listed in Tables 1–3.

Analytic Plan

We used frequencies to characterize children's involvement, provider facilitation, and parent facilitation in the consent sessions. Due to the skewed distributions of these variables, we created dichotomous variables for presence versus absence of any child communication (Child Involvement), parent facilitation of child involvement (Parent Facilitate), and provider facilitation of child involvement (Provider Facilitate) during the session. We used Fisher's exact tests to test the associations among child involvement, parent facilitation, and provider facilitation. We used independent samples t-tests to test the associations of children's involvement, provider facilitation, and parent facilitation with child age and duration of consent session. Given that child assent was waived for children in the neurodevelopmental disabilities cohort, we omitted this cohort and reran the Fisher's exact tests and independent samples t-tests. The pattern of findings was consistent with those from the complete sample, so the results that follow reflect data based on the complete sample.

Results

Participants

Two hundred and thirteen families were approached for participation in the study, and 191 (90%) agreed. Of these, 145 (76%) had an audiotape of the consent discussion. We had funding to code the first 50 audiotapes we received for probands ages 8–21 years. Because communication patterns may be different when interacting with probands who are assenting versus probands who are consenting for themselves, we focused on the 44 sessions in which the youth was age 8–17 years for this analysis. Information about participant and consent session characteristics is in Table 4.

Descriptive Findings Related to Children's Involvement

In 75% (n = 33) of the consent sessions, the child verbally participated in some way. Of the 33 children who participated in some way, 82% (n = 27) made comments related to his/her health or study participation, 79% (n = 26) made a simple response (yes, no, or okay), and 49% (n = 16) made comments unrelated to his/her health or study participation (i.e., chit-chat). Of the 27 sessions in which the child made comments related to his/her health or study

participation, the most frequent types of comments were asking a question or expressing a concern (n = 20, 74%), expressing an opinion about study participation or secondary findings (n = 14, 52%), and providing information or an opinion about something other than research participation or secondary findings (n = 14, 52%)(Table 1).

We further explored children's questions and concerns about study participation (Table 2). For the 20 participants who asked a question or expressed a concern, there were a total of 58 questions/concerns. Of these 58 questions/concerns, most related to practical issues/procedures (n= 26 questions/concerns, 45%) and the consent form or brochures (n= 17 questions/concerns, 29%). Questions/concerns about practical issues/procedures had to do with topics such as inclusion of results in the medical record, accuracy of the results, when the results would be available, when the blood sample would be drawn, how many vials of blood would be drawn, incentives for study participation, whether or not the audio-recorder was on, and whether the consent discussion was over yet. Although infrequent, questions/concerns related to risks or negative consequences mostly related to secondary findings, such as anxiety about the results and concern that a secondary finding would result in a limitation on the child's activities. Table 2 provides selected quotes from each of the content categories.

In 73% (n = 32) of consent sessions, the provider attempted to facilitate the child's involvement. Of these sessions, the most frequent types of facilitation were soliciting questions (occurring in n = 20 sessions, 63%), asking for the child's opinion (n = 18, 56%), and checking for child understanding (n = 16, 50%)(Table 3). In 43% (n = 19) of the consent sessions, the parent attempted to facilitate the child's involvement. Of these sessions, the most frequent types of facilitation were asking for the child's opinion (n = 8, 42%) and soliciting questions (n = 7, 37%)(Table 3).

In 27% (n = 12) of the 44 consent sessions, there were no instances of either provider or parent facilitation of the child's involvement.

Table 5 shows the variation in the frequencies of child involvement and provider and parent facilitation by condition cohort. However, because cohorts were almost synonymous with study physician (e.g., there was one study physician for all children in the cardiac arrhythmia cohort), it is impossible to conclude whether the variation is due to the specific provider who were involved in the consent process or the condition cohort. Therefore, we did not analyze this apparent variation further.

Associations of Children's Involvement with Parent and Provider Facilitation

We used Fisher's exact test to examine the associations among Child Involvement, Parent Facilitate, and Provider Facilitate. Children's involvement in the session was more likely if the provider attempted to facilitate such involvement (p = .000). When the provider attempted to facilitate the child's involvement, 97% of children participated. In contrast, when providers did not attempt to facilitate, only 17% of children participated. Children's involvement in the session was also more likely when parents attempted to facilitate such involvement (p = .013). When the parent attempted to facilitate the child's involvement, 95% of children participated. When parents did not attempt to facilitate, only 60% of children

participated. Finally, parent facilitation of the child's involvement was more likely when the provider also facilitated the child's involvement (p = .000). When the provider attempted to facilitate the child's involvement, 59% of parents also attempted to facilitate the child's involvement. When providers did not attempt to facilitate, no parents attempted to facilitate.

Given that Provider Facilitate was highly associated with the likelihood of child involvement, we explored whether specific provider facilitation categories drove this association. Asking for the child's opinion (p = .001), checking for the child's understanding (p = .003), soliciting questions (p = .000), and chit-chat (p = .008) were each associated with greater likelihood of child involvement. For each of these facilitation categories, child involvement was less likely when the behavior was absent compared to when the behavior was present (e.g., when the provider asked for the child's opinion, 100% of the children participated, but when the provider did not ask for the child's opinion, only 58% of children participated). Asking the child a question, asking for the child's signature, checking for self-understanding of what the child has said, and other forms of facilitation were not associated with likelihood of child involvement.

Associations of Children's Involvement with Age and Duration of Consent Session

Child age was older when Child Involvement was present versus absent ($t(41.67) = -6.90$, $p < .0001$; 14.00 vs. 9.84), when Parent Facilitate was present versus absent ($t(42) = -3.41$, $p = .001$; 14.63 vs. 11.69), and when Provider Facilitate was present versus absent ($t(41.96) = -7.53$, $p < .0001$; 14.17 vs. 9.74).

The duration of the informed consent session was longer when Parent Facilitate was present versus absent ($t(42) = -2.65$, $p = .011$; 48.63 vs. 35.36 minutes) and when Provider Facilitate was present versus absent ($t(42) = -2.19$, $p = .034$; 44.5 vs. 32 minutes). Duration of the session was not associated with Child Involvement.

Discussion

The results of this observational study of informed consent for pediatric exome research indicated that the majority of providers attempted to facilitate child participation and when they did, it was most frequently in the form of soliciting questions, asking for the child's opinion, and checking for child understanding. Furthermore, provider facilitation was strongly associated with the likelihood of child verbal participation in the consent session. Specific forms of provider facilitation that were associated with child participation included asking for the child's opinion, checking for the child's understanding, soliciting questions, and chit-chat. These types of behaviors focus on the process and relational aspects of decision making, which may be more relevant and/or engaging to children than disclosure of vast amounts of information related to the study protocol. Furthermore, these behaviors show respect for children by situating the child as central to the decision making process.

Most children participated in the consent sessions in some way, including chit-chat, simple responses to provider or parent questions/comments, and voicing concerns or questions about the research study. When children asked a question or expressed a concern, it most frequently had to do with practical issues or the procedures of the research study. A minority

of children's questions/concerns related to the risks of participation. This is not surprising, considering that research risks are less frequently understood by children compared to more concrete aspects of participation, such as duration of involvement in the study and how to withdraw from the study (Miller, Drotar, & Kodish, 2004). While the uncertainty and potential long-term implications of exome sequencing may be salient for parents, children appear to have different concerns, which is consistent with prior focus group research regarding exome sequencing (Levenseller et al., 2013).

In a sizable minority of the consent sessions (27%), neither providers nor parents attempted to facilitate the child's involvement in the session. Furthermore, 25% of the sessions contained no child verbal involvement of any kind. These sessions were not limited to participants from the hearing loss cohort or for whom the assent requirement was waived, so it is not clear what is driving the lack of provider/parent facilitation and child involvement. One potential reason for lack of provider or parent facilitation is that involving children in the consent discussion takes more time. On average, sessions in which the provider or parent attempted to facilitate the child's involvement were about 14 minutes longer than sessions in which they did not. Informed consent sessions typically occurred in conjunction with clinical care visits, during which a physical exam was conducted and issues relevant to clinical care were discussed. If providers, in particular, are aware of the additional time it takes to engage children in discussions about research, they may be reluctant to attempt to increase their involvement in the face of competing demands on their time.

Another potential explanation for lack of parent or provider facilitation or child involvement is the child's age; consistent with prior research (Miller, Baker, Leek, Drotar, & Kodish, 2014), child age was younger in sessions in which child involvement, parent facilitation, and provider facilitation were absent. Younger children may lack the maturity and/or communication skills to speak up during medical encounters, especially when multiple authority figures are present, and/or they may be accustomed to parents assuming primary responsibility for decision making. In addition, providers may equate the assent requirement with disclosing information and obtaining a signature on the consent form. As such, they may be unaware of strategies to facilitate children in the decision making process, especially in the context of decision making about opting to learn about secondary findings. As Clayton (Clayton, 2015) notes, the American College of Medical Genetics and Genomics does not provide guidance regarding ways to engage minors in decisions about secondary findings. Further, participants may need to provide consent for ongoing analysis of their samples after they turn 18 (Brothers et al., 2016). As such, the initial process of decision making about research participation and opting to receive secondary findings is especially important.

Limitations of this study include the small sample size and that only a subset of available audiotapes was coded. We relied on audiotapes of the sessions and, therefore, were unable to code non-verbal communication. The results were purely descriptive and we did not measure outcomes of children's involvement. Furthermore, the results may be biased in that we only included individuals who consented to enroll in the exome sequencing research and not those who declined. In addition, the sample was of limited diversity with respect to child race and parental education status; as such, the results cannot be generalized to non-White,

less educated samples. Finally, it is possible that discussions about research participation, including decision making about secondary findings, may have occurred outside of the consent sessions that were analyzed here.

Best Practices

Our data suggest that there is room for improvement with respect to children's involvement in decisions about exome sequencing research and disclosure of secondary findings. Providers and other research personnel should consider specific communication strategies to increase the likelihood of children's verbal participation in the consent session, which include asking for the child's opinion, checking for the child's understanding, soliciting questions, and chit-chat. Furthermore, the information that is provided to children during the assent process for exome sequencing research should start with what is most salient to children, which included procedures and practical issues related to study participation. Study personnel can address more complex or abstract topics, such as purpose and potential risks, with more mature children.

Research Agenda

Future research is needed to identify reasons for lack of child involvement and lack of provider facilitation in a subset of sessions, using a prospective design and multi-method assessment strategy, including both observational and self-report data. An understanding of the factors that drive child, provider, and parent communication patterns about both exome sequencing and disclosure of secondary findings will facilitate the development of intervention strategies to enhance respect for children during the decision making process. Additional research is also needed to document outcomes of child involvement. Although obtaining assent is important in its own right, the assent process may also increase children's satisfaction with the decision making process, improve their understanding of the research, and increase decision making self-efficacy.

Educational Implications

It is critical for investigators and individuals conducting informed consent with families to understand that there are multiple strategies to show respect for children and encourage their active involvement in consent discussions. IRBs should consider educational initiatives, such as training sessions and toolkits, to increase awareness and provide specific guidance regarding communication strategies to increase child participation in informed consent sessions in general, and in decision making about exome sequencing research with disclosure of secondary findings in particular.

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Table 1

Frequencies of Child Involvement Categories

	Child Involvement (N = 33) N (%)
Chit-chat	16 (49%)
Simple response (yes, no, okay)	26 (79%)
Study/health-related comments	27 (82%)
Sub-categories for study/health-related comments (N = 27)	
Ask question or express concern	20 (74%)
Express opinion about study or secondary findings	14 (52%)
Provide information	14 (52%)
Check self-understanding of what provider/parent said	4 (15%)
Answer “quiz” question posed by provider/parent	4 (15%)
Make a joke	3 (11%)
Other	3 (11%)

Note: Values indicate number and percentage of consent discussions in which the behavior was present.

Table 2

Frequencies and Examples of Children's Questions and Concerns (N = 58 questions/concerns)

	N (%)	Example
Practical issues/procedures	26 (45%)	How many blood tubes do we get taken? (9 y.o. male) Are they going to do it now, are they going to do the testing now? (12 y.o. male)
Consent form/brochures	17 (29%)	Where do I sign? (15 y.o. male)
Risks/negative consequences	7 (12%)	There's nothing to worry about, right? They're not going to be like, horrible results, or anything. Like if it was some, like, life-threatening, I'd already have it, right? (16 y.o. female) You're gonna find something [referring to secondary findings] and I'm not going to be able to play soccer again (17 y.o. male) I don't like getting my blood taken. (9 y.o. male)
Other	4 (7%)	Mother: So we're pretty okay dealing with the level of uncertainty so far, so." Provider: Okay. Proband: The carrier one, too? (17 y.o. female)
Genetics	1 (2%)	You say carrier, does that rule out homozygous recessive? (15 y.o. male)
Sample/design	1 (2%)	What are like the ages of people getting tested, like me? (17 y.o. female)
Benefits/positive consequences	1 (2%)	Am I able to say that I helped map the human genome by being, like, a participant in this? (15 y.o. male)
Medical/family history	1 (2%)	What's dyslexic [referring to family member]? (16 y.o. female)
Research rights	1 (2%)	Do I have to do it [participate in the study]? (9 y.o. male)

Note: Values indicate number and percentage of questions/concerns from each category, out of 58 total questions/concerns

Table 3

Frequencies of Provider and Parent Facilitation Categories

	Provider Facilitate (N = 32) N (%)	**Parent Facilitate (N = 19) N (%)**
Solicit questions	20 (63%)	7 (37%)
Ask for child's opinion	18 (56%)	8 (42%)
Check that child understands	16 (50%)	3 (16%)
Chit-chat	15 (47%)	N/A
Asks for signature	12 (38%)	6 (32%)
Check self-understanding of what child has said	8 (25%)	0
Other	6 (19%)	4 (21%)
Asks questions about medical/family history	2 (6%)	0

Note: Values indicate number and percentage of consent discussions in which the behavior was present.

Table 4

Participant and Consent Session Characteristics

	Mean (SD, range) or N (%)
Child age (years)	12.96 (3.17, 8–17)
Child sex	
Male	28 (64%)
Female	16 (36%)
Child race	
White	35 (80%)
More than one race	4 (9%)
African-American	3 (7%)
Asian	1 (2%)
Unknown	1 (2%)
Disease cohort	
Cardiac arrhythmia or cardiomyopathy	22 (50%)
Neurodevelopmental disorder	7 (16%)
Non-syndromic hearing loss	6 (14%)
Nuclear encoded mitochondrial disease	5 (11%)
Platelet function disorder	4 (9%)
Parent marital status	
Married/living as married	37 (84%)
Divorced	3 (7%)
Widowed	1 (2%)
Missing	3 (7%)
Parent educational attainment	
Completed high school	4 (9%)
Vocational/technical	4 (9%)
Some college	5 (11%)
College	14 (32%)
Post-graduate	14 (32%)
Missing	3 (7%)
Visit type	
Initial clinical visit	30 (68%)
Follow-up clinical visit	10 (23%)
Research visit	4 (9%)
Clinician/study personnel present during consent session	
Genetic counselor	31 (70%)
Physician	21 (48%)

	Mean (SD, range) or N (%)
Nurse	18 (41%)
Study coordinator	2 (5%)
Number of clinicians/study personnel present during consent session	1.64 (0.72, 1–3)
Duration of consent session (minutes)	41.09 (17.56, 8–90)

Table 5

Frequencies of Provider and Parent Facilitation and Child Involvement by Condition Cohort

Cohort	Provider Facilitate N (%)	Parent Facilitate N (%)	Child Involvement N (%)
Neurodevelopmental disorder (n = 7)	3 (43%)	3 (43%)	4 (57%)
Non-syndromic hearing loss (n = 6)	2 (33%)	0	2 (33%)
Cardiac arrhythmia/cardiomyopathy (n = 22)	21 (95%)	11 (50%)	16 (73%)
Platelet function disorder (n = 4)	3 (75%)	3 (75%)	2 (50%)
Nuclear encoded mitochondrial disease (n = 5)	3 (60%)	2 (40%)	3 (60%)

Note: Values indicate number and percentage of consent discussions from that condition cohort in which the behavior was present