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Shortened consent forms for genome-wide sequencing: Parent and provider perspectives

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Abstract

Background: Consent forms for exome and/or genome sequencing, collectively called genome-wide sequencing (GWS), frequently contain detailed information on complex topics such as sequencing analysis and incidental findings. Considering recent endeavors by the health care community to simplify GWS consent forms, it is important to gain stakeholders' perspectives on the content, length, and use of consent forms.

Methods: Thematic analysis was conducted on data obtained from focus groups with two participant cohorts: parents who previously provided consent for trio-based GWS as part of the translational pediatric GWS CAUSES Study, and genetic health care providers (HCP) who provide pre-test counseling for GWS.

Results: Genetic HCP indicated that consent forms cannot replace pre-test counseling, and as such, a simplified consent form focusing on the implications of GWS would be beneficial to both patients and HCP. Although parents' primary concerns varied when considering GWS, they all highly valued information. Parents also indicated the need for community and support after the return of GWS results. Both participant cohorts recommended that consent forms be available online and include an appendix for supplementary information.

Conclusion: It is important to include both parents and HCP in the design of GWS consent forms, and also, to help connect families who have a shared diagnosis after the post-test counseling session.

KEYWORDS

exome sequencing, genetic counseling, genome sequencing, genome-wide sequencing, informed consent

1 | INTRODUCTION

Informed consent (IC) is a required component of most clinical and all research genetic testing. Guidelines for the ethical conduct of research in Canada and the Code of Federal Regulations in the United States require research participants receive all necessary information before making an informed decision regarding study participation (Basic HHS Policy for Protection of Human Research Subjects, 2018; Canadian Institutes of Health Research, Natural Sciences and Engineering Research Council of Canada, and Social Sciences and Humanities Research Council, 2018). In general, research consent forms have increased in length, progressively containing a higher level of detail and a more complete description of study risks (Albala, Doyle, & Appelbaum, 2010; Beardsley, Jefford, & Mileshkin, 2007). Studies involving exome and genome sequencing, collectively referred to as genome-wide sequencing (GWS), need

to provide potential participants information beyond the standard elements of research, such as the potential risks and benefits of participating. It is recommended that individuals being consented for GWS be informed about: the limitations of sequencing, the possible types of results and incidental findings, the implications of testing for family members, the possible collection of photographic data, the sharing of data across centers or databases, and if and how patients can re-contact the study team for updated variant interpretation (ACMG Board of Directors, 2013, 2015; Burke & Clarke, 2016; Dheensa et al., 2018; Elliott & Friedman, 2018; Nguyen et al., 2019). Consequently, GWS consent forms are long, complex, and often fail to meet the recommended grade 8 reading level (Fowler, Saunders, & Hoffman, 2017; Henderson et al., 2014; Niemiec, Vears, Borry, & Howard, 2017). The additional considerations for GWS can create challenges in obtaining informed consent (Grady, 2015; Tomlinson et al., 2016). It has been questioned whether it is possible for patients to give IC for GWS studies, as the large amount of information being conveyed could cause overwhelm and a subsequent decrease in patient decision-making capacity (Bester, Cole, & Kodish, 2016).

There has been an effort made by the health care community to simplify the consenting process. Patient knowledge has either increased or remained the same with simplified, shortened consent forms for GWS, biobanking, and clinical trials (Beskow, Lin, Dombeck, Gao, & Weinfurt, 2017; Enama et al., 2012; Kim & Kim, 2015; Kost, Poppel, & Coller, 2017; Turbitt et al., 2018). While knowledge is a large component of IC, past research does not provide insight into the patient experience, including patient satisfaction or usefulness of the consent form during and after the IC process. Therefore, health care providers (HCP) could be removing information important to patients and families (that does not affect knowledge) when attempting to simplify the consenting process. Alternately, shortened consent forms could be beneficial in reducing patient and family burden by removing extra information irrelevant to obtaining IC.

Research has explored parental perceptions or expectations of different aspects of GWS, including preferences for return of results and the psychosocial impact of sequencing results (Krabbenborg et al., 2016; Rigter et al., 2014; Sapp et al., 2013). Li et al. (2016) interviewed 15 parents who provided consent for GWS on behalf of their child, and found that despite most participants feeling that they received "enough" information, there were self-perceived gaps in knowledge about GWS. Participants valued trust and communication with HCP during and after the consenting process. Participants also suggested providing educational information to parents to take home after the pre-test counseling session. However, to the best of our knowledge, patient perspectives specifically on consent forms for GWS have not yet been studied. Before further efforts are undertaken to simplify consent forms, it is important to explore insights from patients and their families.

In this study, we gathered opinions and feedback through focus groups from parents who provided informed consent for themselves and on behalf of their child for trio-based GWS, and from genetic HCP, (including medical geneticists, genetic counselors, and clinician researchers), on an example of a standard length and a shortened consent form for GWS. The sample consent forms were based on the consent form used by the Clinical Assessment of the Utility of Sequencing and Evaluation as a Service (CAUSES) Study based at BC Children's and Women's Hospitals in Vancouver, British Columbia. The CAUSES Study was a translational research study that used trio-based GWS for children with suspected, but undiagnosed, genetic disorders (Dragojlovic et al., 2018, 2019; Elliott et al., 2018; Inglese, Elliott, CAUSES Study, & Lehman, 2019; Smith et al., 2019). At the time of the CAUSES Study, GWS funding through the provincial Medical Services Plan in British Columbia was limited, and many families would not have had access to GWS without participating in research. Parent participants (PP) and genetic health care provider participants (GHP) were included in the current study (in separate groupings) so that perspectives could be compared to identify similarities and differences in opinions.

2 | MATERIALS

2.1 | Sample consent forms

The CAUSES Study assessed the utility of GWS as a diagnostic tool and included genetic counseling and health economic research. As such, the consent form contained information not commonly found in consent forms for GWS. The original CAUSES Study consent form was modified to create an example "standard length" consent form (12 pages), and then further modified to create an example of a "shortened" consent form (7 pages, 45% fewer words) by removing detailed explanations, but leaving relevant information about GWS and information required by the Research Ethics Board (REB). Relevant information for IC for GWS was determined using the guidelines provided by Burke and Clarke (2016). Participants received the two sample consent forms prior to attending the focus group.

3 | METHODS

3.1 | Ethical compliance

This study was approved by the University of British Columbia/Children's and Women's Health Centre of British

Columbia REB (H17-03213). Participants were approached for participation and provided informed consent following the REB approved protocol. Written IC was obtained via email or in-person prior to the start of the focus group. Data collection took place at BC Children's and Women's Hospitals. Prior to the development of this study, we met with the Chair of the REB to discuss the growing length and complexity of consent forms for GWS studies, which was also a concern to REB members.

3.2 | Participants

We recruited parent participants (PP) from the cohort of parents who previously provided IC for trio-based GWS as part of the CAUSES Study, and who had additionally consented to be approached for follow-up research. The genetic health care provider participant (GHP) group represented a diverse group of clinicians and researchers and was recruited from the Provincial Medical Genetics Program at BC Children's and Women's Hospitals, the BC Hereditary Cancer Program, and the BC Inherited Arrhythmia Program. These three programs provide genetic health services to the population of British Columbia.

3.3 | Data collection and analysis

Focus groups were selected as the method of data collection as they allow for participants to share their own perspectives as well as to discuss their opinions with peers (Morgan, King, & Krueger, 1998). Focus groups were led by E. C. H. and overseen by A. M. E. Two focus groups were conducted with the PP and one focus group with the GHP about the readability and content of the two sample consent forms. As well, participants in all three groups were asked about their use of the consent forms both during and after the consenting process. Focus groups were semi-structured and guided by a predetermined list of questions designed using the Focus Group Kit (Morgan et al., 1998) to facilitate open ended conversation. Each focus group was 90 min in length and aimed to consist of five to ten participants.

Non-identifiable demographic information was collected at the start of each focus group. Variables collected from GHP included age, sex, ethnicity, profession, years practicing, and field of practice. Variables collected from PP included age, sex, ethnicity, as well as variables related to their child with a suspected genetic condition including genomic diagnosis status, age at genomic diagnosis, and the number of years their child was suspected to have a genetic condition.

Focus group data was analyzed through full verbatim transcription, and each transcript was checked against the corresponding recording. As perspectives on consent forms is a previously understudied area, an inductive approach was chosen for thematic analysis of transcripts (Braun & Clarke, 2006). Extracts containing participant perspectives and opinions were identified in the transcript and coded. As analysis progressed, early codes were revised and/or merged with other codes where necessary. Codes were then collated, and themes were identified for each focus group. Codes and themes from the parent participant focus groups were analyzed independently, and then together, to identify themes that were representative of both groups. Initial analysis of the complete data set was done by E. C. H. The transcripts from both focus groups were independently coded by both E. C. H. and A. M. E. Discrepancies in codes and themes were resolved and initial coding was revised where necessary. Themes were validated by verifying each theme was representative of the corresponding focus group and checked for appropriate fit with coded extracts.

TABLE 1 Demographics of genetic health care provider participants

	Participants ($n = 7$)	Percentage
Sex		
Female	6	86
Male	1	14
Age, years		
Median	48	
Range	32–61	
Ancestry		
European	6	86
East Asian	1	14
Profession		
Genetic Counselor	5	72
Clinical Geneticist	1	14
Clinician Researcher	1	14
Years practicing		
Median	19	
Range	3.5–35	
Field of practice		
Clinical	3	43
Research	3	43
Both	1	14

4 | RESULTS

4.1 | Focus group with Genetic Health Care Provider Participants (GHP)

Eight GHP agreed to participate and seven attended the focus group. The focus group was comprised of five genetic

counselors, one clinical geneticist, and one clinician researcher with experience in the field ranging from 3.5 to 35 years (Table 1). Thematic analysis of the data from the focus group with GHP reached saturation.

4.1.1 | Theme 1: Consent forms should focus on implications of GWS

During the focus group, GHP indicated that the *implications* of GWS were the most relevant considerations for families when deciding whether or not to undergo GWS. They considered the implications of GWS to be any aspect of testing that could impact the family during the study, as well as after the counseling session for return of results. Specifically, the possible results from GWS, the potential risks of GWS, and the study procedures were all seen as impactful on families by GHP. It was also noted by GHP that families often ask questions about these topics during the consenting process, and therefore were assumed to be important to families as well. GHP reached a consensus that possible outcomes of GWS (a positive, negative, or variant of uncertain significance result, as well as the possibility of receiving incidental findings) were of primary importance to discuss with a family undergoing trio-based GWS. In addition, a description of the types of genetic conditions not being purposefully assessed in the GWS study should be included in the consent form. GHP emphasized that they would convey the potential for uncertainty with GWS results and the potential for variant reclassification in the future to the family when discussing possible outcomes. Providing clear information on study procedures, such as the reporting of incidental findings, was seen as potentially having a positive impact by decreasing ambiguity for families.

> *"I think for me the most important thing I try and get across is that this is research, and this is new stuff, and that results may be fuzzy… What we say is our best guess today and may not be our best guess tomorrow, or next year…"– GHP5*

> *"It was an interesting question about returning [incidental findings] for actionable conditions in childhood on their child, and then…conditions that were actionable as adults. And [the parents] said 'well if it's actionable as an adult but you find it in him as a child, can he come back as ask for his results, once he is an adult?' … So to have a clear yes or no for them is helpful, rather than this, kind of, vagaries that could carry on for a long time."– GHP6*

It was thought the explanations in the consent form of complicated subjects, such as the GWS technology and privacy risks, should be reduced in length, and that the information provided on these topics should outline the potential impact to the family. It was recommended that a broad description of GWS be given, with comparisons to targeted genetic tests, to demonstrate the larger scope of testing.

> *"I think the technology and analysis is less important than the implications of what that means for them… the scope of the possible results is what would be meaningful for them, not how they're getting that scope of result." – GHP3*

However, continuing to provide an outline of the analysis of data and potential risks to privacy was seen as valuable for building trust and maintaining transparency"*… in terms of us [as] researchers being upfront with the research participants about how much care we are taking in storing their data,"* (GHP1).

4.1.2 | Theme 2: Consent forms cannot replace pre-test counseling

GHP made distinctions between the purpose of a consent form and the role of pre-test counseling in the IC process. Participants indicated consent forms should provide the minimum information required for informed dissent or consent to participate in a GWS study. Pre-test counseling was seen to provide patient-centered care and a discussion on GWS that can expand on the considerations important to each family at an appropriate resolution of information.

> *"There is a certain level of detail that everybody should know to sign the paper, but then beyond that, some people will be much more concerned or interested in detail than others. …I think it's helpful to gauge that and give information. I don't know if every detail needs to be included [in the consent form]." – GHP3*

Having a personalized conversation also provided space for GHP to resolve any misconceptions about GWS that might be held by patients, such as the expectation that research GWS would have a comparable turn-around-time to clinical GWS. GHP noted that patients often are confused between clinical and research GWS. GHP perceived the potential implications, risks, and benefits as being too vast a subject to be adequately explained in a consent form. It was unclear to GHP if the information in the consent form was sufficient to impart understanding to families.

"I would say even with this [consent form] you talk about incidental findings and implications and you don't know if potential implications are fully grasped, by a lot of people." – GHP7

"One of the most challenging things …is explaining a theoretical risk, or a very low theoretical risk of a privacy breach to the extent that we think someone might need to understand it because you can never know how much someone needs to know to understand it until they ask detailed questions. …and it is difficult to quantify given the shifts in technology that have occurred over time."– GHP1

GHP described situations where there could be barriers to IC if patients were only provided with a consent form. Many GHP felt that their patients did not read the consent form and saw pre-test counseling as a way to ensure that patients could discuss GWS and provide IC. It also was indicated that some environments, such as the neonatal intensive care unit (NICU), might only permit HCP to obtain IC and not allow for effective pre-test counseling.

"And again, the counseling is really – if you've had a good counseling session, they should understand generally what's in there, whether they actually read it, and then they have it sort of to refer back to, if they want to." – GHP3

"But often times, when we're doing trio exomes we are in the NICU, and there are beeps going on, and you're not having a counseling session, right?" – GHP8

4.1.3 | Theme 3: Simplified consent forms are seen as beneficial

All GHP preferred the length of the shortened sample consent form. However, GHP did not see shortening consent forms as being sufficient to positively impact the consenting process. GHP saw lowering the reading level, reducing the density of information, and changing the format of consent forms, as additional improvements that would be beneficial for both providers and patients, and necessary for responsible practice.

"I think shortening and bullet points could really help, like, it's kind of unethical to have a 12-page consent form."– GHP5

A simplified consent form was seen as beneficial by GHP as administering it would require less time both before and during the pre-test counseling session. GHP thought that the shortened and simplified consent form would allow HCP to more quickly and clearly understand the requirements and procedures of a GWS study, and in turn be able to convey this information to patients. Additionally, GHP stated they would be more likely to use a simplified consent form in a clinic appointment.

"I would be, I think, more likely to use the short consent form, …then and there, …in the actual session. And more likely to provide a form of the length of the standard form essentially at the end of the session and say, 'get back to us about this.'"– GHP1

"…it needs to be quick and dirty for me so that I can make it quick and dirty for the patient. … it's about access for me as well as access for the patient. …I don't want to go through 12 pages of yet another consent form …if it can be done in three. And in bullet points, highlights."– GHP7

This increased efficiency was important to providers. GHP described wanting all components of a study, required and optional, in a single, simplified consent form even at the expense of patient understanding. One GHP described their past experience consenting for a GWS study (unrelated to the CAUSES Study) that, at times, involved tissue collection.

"…we had a very broad [consent form from an external study] that covered a few different cohorts. Most people were having a blood sample, but [the consent form] mentions skin biopsies, bone marrow, and a few other things, and everybody went, 'oh my God, does my child have to have a skin biopsy?' And the answer was always no, it was just a really broad consent in case we wanted to do functional studies after the fact. And everybody came back really worried about that and had a lot of questions, um, yeah, every time."– GHP3

GHP identified increased comprehension of study information and increased access to GWS studies as the main benefits to patients. The density and complexity of information in the sample consent forms was seen as potentially detrimental to patients. Comparatively, simplifying and restructuring the consent form was thought to provide better clarity and access to key study components for patients.

"Well actually I think that level of detail is almost frightening."

"Yeah, it's off-putting, and a little bit anxiety provoking."– GHP2 & GHP6

"I mean the real risks of a genetic study like this are the privacy risks or uncovering information that the family wasn't previously prepared for. It would be nice to see those in bullet point format."– GHP1

Simplified consent forms were also seen as having the potential to increase health equity by helping serve patients who do not read English as their primary language.

"…the shorter [the consent form] is, the easier it is to translate into other languages. So that's another really important thing about equity."– GHP5

4.2 | Focus group with Parent Participants (PP)

Eleven parents agreed to participate and eight attended the focus groups (Table 2). Two couples attended the focus groups; therefore, the PP cohort consisted of eight parents from six families. For four families, their child received a genomic diagnosis from GWS (a positive result and a variant of uncertain significance suspected to be causative). For two families, their child received a negative result from GWS. Thematic analysis of the data from the focus group with PP reached saturation.

TABLE 2 Demographics of parent participants and their children with a suspected genetic condition

	Participants (n = 8)	Percentage
Sex		
Female	5	60
Male	3	40
Age, years		
Median	37.5	
Range	36–47	
Ancestry		
European	6	75
South American	1	13
Other	2	25
	Children of Participants (n = 6)	**Percentage**
Sequencing result		
Positive	3	50
VUS	1	17
Negative	2	33
Age at genomic diagnosis,[a] years		
Median	15.5	
Range	1–20	
Diagnostic odyssey,[b] years		
Median	8.7	
Range	<1–19	

Abbreviations: VUS, variant of uncertain significance.

[a]Positive result and VUS considered a diagnosis.

[b]Refers to the time between the age at which a genetic condition was suspected and the age at genomic diagnosis or current age (if undiagnosed).

4.2.1 | Theme 1: Importance of Information

PP placed a high importance on information they obtained from the consent form, through communication with HCP, and/or through independent research online. Although PP indicated they had varying informational needs, all parents indicated that the shortened sample consent form had insufficient detail and clarity. One PP felt it was important that detailed information be provided through the consent form, as it was unlikely that parents would have prior knowledge about GWS. PP felt that it was necessary for consent forms to provide a more detailed description on the potential for incidental findings and the potential risks associated with GWS studies (most predominantly the risk for genetic discrimination by insurance companies). Many of the PP asked about the potential impact of sequencing results and incidental findings on obtaining insurance at their pre-test counseling appointment. One participant was concerned enough to independently research the current Canadian legislation protecting individuals against genetic discrimination (Genetic Non-Discrimination Act, Bill S-201). PP indicated information on the implications of GWS on obtaining insurance in the future would be helpful to include in the consent form.

"For me, it is that confidentiality that it's not going to, um, wreck his chances of getting… private medical insurance when he's old enough. You know when he's got a job or a career that he won't be denied that. … That was important to me."–PP6

PP viewed the consent form as a resource for information. Some PP described re-reading the consent form during their participation in the CAUSES Study (but after providing IC). It was important to PP that the consent form provide a clear study rationale and a clear description of study procedures, such as the timeline and the requirements for participation. Although, not all PP utilized the consent form in the same way, the information in the consent form was empowering

both during the study and after the return of GWS results. Almost all PP used the information in the consent form as a basis for their own independent research into GWS before giving consent to participate in the study. The consent forms provided information outside of the pre-test counseling session which allowed PP to process the information at their own pace.

> *"For myself, yes, I probably looked at it, probably five times. …speaking of my own experience, when I'm overwhelmed, I don't retain everything, um so, when I speak through it, take a couple of days to digest it, um, going back because maybe I have somebody else's questions that I didn't think about and then I can go back and reference, it was really helpful." – PP4*

> *"I like the longer version, 'cause then I get more of an idea of what it's about, and then I can look it up or choose to look it up. Yeah, so, like I want to know everything."– PP8*

We observed that a key time point that some PP referenced the consent form was before they returned to clinic for the results counseling session.

> *"… I went back over [the consent form] once just before, when we knew we were getting our … results, …right where it steps through the results and the process we were about to go through … it was kind of like to refresh, like, what we were about to deal with."– PP2*

After the results session, some PP felt the information in the consent form gave them the option of pursuing investigations external to the CAUSES Study with other HCP. These PP advocated for detailed information in the consent form.

> *"So as much [information] as possible (laughs), even when it's too much for us as parents, it's just good to have that information in my hands, ah, to do with what I would like."– PP3*

Many PP felt it was important to be connected with members of the study team, both during and after the pre-test counseling session, to have their questions addressed. One parent described being comfortable with participating in the GWS study because of their trust in the study team. The explanation of the study during the pre-test counseling session was described as being especially helpful by one parent.

> *"When [my daughter] and I came to meet with the [genetic counselor]… she was fantastic how she explained it. So even [my daughter] got it, like, it was so simple."– PP8*

4.2.2 | Theme 2: Support after return of results

The PP indicated that information provided at pre- or post-test counseling sessions, and in the consent form, to help guide families after the completion of the study could be valuable. One parent described questioning "*what's next?*" and being directionless after their results session. PP were interested in receiving direction to trusted resources as well as methods to connect with other families. It was reassuring for another PP to read in the consent form that if an incidental finding was identified then support from an HCP, in this case, an appointment with a genetic counselor, would be available regarding that finding.

> *"And I think [the consent form] compartmentalized that and it was perfect because you could shelf it and if it happened, then we would look back, and then say, oh look see it says here that you can make an appointment, that's what we'll do." – PP3*

PP expressed altruism and described helping other families reach a diagnosis in the future as a motivating factor to participate in the GWS study. A component of the consent form for the CAUSES Study was an option to allow anonymized data to be shared, and PP viewed opting in as a way to contribute to the greater understanding of genetic disorders. Families and individuals who could share their lived experience were seen as another source of guidance and a potential source of support by PP.

> *You know, even a feeling, a sense of community, right? …there's so many parents, like us, who are going through this… just to let us know otherpeople are involved, even in the consent [form] so that we're not breaking ground. – PP9*

> *"You've done the genetic counseling, where can you reach out to like-minded or other families that …are going through the same journey. … we may never meet face to face, but yet here I found someone across the world that has the same analysis that my [child] has. – PP4*

> *"'Cause I would love that so much, if I could have an answer… for [my son] to have someone who's like 'I get it,' like, that would be amazing… that connection would be so cool."– PP6*

One PP recommended that the consent form state that parents can ask their genetic counselor at the results session about resources, such as support groups. PP seemed to have felt comforted by the shared experience of participating in a study that included 500 families with suspected genetic disorders, and privileged to have had access to GWS.

4.2.3 | Theme 3: Parental priorities are influenced by past experiences and values

Across both focus groups, PP had different priorities when considering trio-based GWS that were guided by their values or past experiences. The information PP thought should be emphasized in the consent form reflected their priorities and concerns. Despite some PP having a shared family experience with their spouse, each parent had different informational needs regarding GWS. One PP reported a motivation for testing was to find a diagnosis for their child and that the considerations of GWS that focused on the parents, such as the reporting of incidental findings (identified in the parents), were less important and therefore did not need as much explanation in the consent form. A number of PP experienced a long diagnostic odyssey with their child and the prospect of finding a diagnosis was both exciting and motivating. Some PP reported that they would "*sign no matter what*" as the chance to find a diagnosis for their child outweighed any potential risks of the study. However, these PP maintained that the consent form should contain comprehensive information about study risks. Additionally, when considering participation in a GWS study, finding a diagnosis was not always reported as their primary concern. PP described the exhaustion and disruption to every-day life caused by frequent medical appointments. It was important for PP to know the direct impact of the GWS study on their child as well as the indirect impact to their family.

> *"I'm guessing in this room everyone has been through lots of testing, and …this is just another thing, so the first thing I look for is what do I gotta do with [my son]? What are you taking? Are you taking blood? Are you taking urine? How long [is the study]? You know, like all those very practical …questions are always what I'll scan for first … and then step by step, what you're expecting from us."– PP3*

Due to a past experience with genetic testing (chromosomal microarray analysis), the biggest concern for one PP was the potential documentation of GWS results into their child's medical record. Information addressing this concern was not provided in the consent form.

> *"…I really needed to know … how, um, the results would be reported in [my child's] medical records, because before we had this done, we had microarray testing done, and it came back with, ah, a variation of unknown significance. …and all of our doctors latched onto it as, like, this is the reason for everything that's going on with your [child]."– PP2*

As discussed previously, concerns around confidentiality, as well as aspirations of finding a community with other families, were important aspects of GWS research that PP recommended be included in the consent form.

4.3 | Perspectives on Consent Forms (GHP and PP)

All GHP and most PP reported that they think improving consent forms is important. During the focus groups, PP discussed that the complexity and length of consent forms were barriers to understanding the information being presented. PP recommended that consent forms be written in lay language with an option for more detailed descriptions. All three participant focus groups independently initiated discussion on restructuring the consent form to include an appendix, which would provide room for supplementary information (such as definitions of terminology), that may be important for decision making in some families but not considered crucial for IC. The PP suggested this alternate format as a way to include more comprehensive information about GWS, while the GHP saw the change in structure as a way to provide a highly simplified consent form before the appendix. PP also acknowledged that some parents may want less information, and that providing a simplified consent form with an appendix could fulfill the needs of both preferences. Participants from all focus groups were interested in an online format, which would allow for easier navigation to appendices and definitions of technical terms, and also in using videos and pictures to convey or receive information. The GHP recommended the use of bullet points, so that key pieces of information could be more accessible in the consent form, and that the amount of REB required text should be minimized. GHP indicated that the main implications of GWS could vary with patient population and this should be reflected in consent forms.

5 | DISCUSSION

This qualitative study is the first to examine parental opinions on consent forms for trio-based GWS in the pediatric setting and adds to the perspectives of HCP on the consenting

process for GWS. GHP regarded a simplified consent form that focused on the implications of GWS, primarily the possible results of GWS, as beneficial to both themselves, through increased utility, and to patients, through increased accessibility of information. PP advocated for consent forms to contain detailed study information. The information PP felt was important to include varied based on past experiences and personal values. However, incidental findings and potential study risks were often raised as concerns by PP. After the return of GWS results, PP wanted guidance and support from the study team and/or other families who shared a similarly lived experience. Additionally, this study provides recommendations from parent and genetic health care provider participants for restructuring consent forms for GWS studies.

Pre-test counseling with a genetic counselor (or other qualified HCP) is not the same as IC and should precede IC for GWS (Elliott & Friedman, 2018). In our study, GHP strongly communicated the necessity of a pre-test counseling session to facilitate informed decision making by patients and seemed to view the consent form as complementary to the session. A study that interviewed genetic counselors and research coordinators experienced in obtaining IC for GWS, found that the pre-test counseling sessions focused on facilitating patient understanding and not on standard elements of consent forms (such as the voluntary nature of research) (Bernhardt et al., 2015). A recent online survey completed by 342 members of the National Society of Genetic Counselors demonstrated counselors prioritize individual patient needs when obtaining informed consent for GWS. In addition, counselors prioritized collaborative decision making, assessing understanding and managing expectations, while the discussion of complex technologies was less likely to be emphasized (Gore, Bridges, Cohen, & Biesecker, 2019). Interestingly, in the current study one GHP indicated the atmosphere of the neonatal intensive care unit (NICU) created a barrier to pre-test counseling when obtaining consent for GWS. Given the high rates of anxiety and depression in parents of neonates undergoing GWS as compared to the general population, the emotional burden on parents, and limited timeframe to make decisions, it is critical to provide effective pre-test counseling in this setting (Diamonstein, 2019; Smith et al., 2019). Parents of newborns receiving rapid trio-based GWS in the NICU were significantly more likely to identify finding a diagnosis as their primary motivation for pursuing GWS and less likely to opt in for return of incidental findings for themselves, as compared to parents of older pediatric patients in the CAUSES Study cohort (Smith et al., 2019). A study by Pereira et al. (2019) found that parents of newborns participating in BabySeq, a GWS sequencing study, were less likely to be concerned about the associated risks, including the potential for insurance discrimination and maintaining privacy of genomic data, when compared to concern held about these issues by their child's primary care physician or pediatrician. Many parents of newborns (68%) eligible for the BabySeq study declined participation in the study due to lack of interest in research. Among parents considering participation in the BabySeq project, the logistics and/or design of the study was most commonly provided as the reason for declining participation (Genetti et al., 2019). It is possible that parents in the NICU will have different information needs for GWS consent forms and future research should explore perspectives from this population.

The opinion that consent forms are too long is widely shared by HCP as evidenced by the research initiatives to shorten consent forms, and the concerns raised about the negative impact of lengthy consent forms on patient understanding for IC (Beardsley et al., 2007; Beskow et al., 2017). Although GHP perceived a simplified consent form as a means to improve patient comprehension, studies on the patient-perceived readability of consent forms written at or above a grade 12 reading level have conflicting results (Manta, Ortiz, Moulton, & Sonnad, 2016; Sommers, Van Staden, & Steffens, 2017). The shared recommendation from the focus group participants in this study, to use a simplified consent form with an appendix, presents a format that addresses the concerns of HCP regarding the length of consent forms and meets the informational needs of parents. The published literature on consent form length and complexity has primarily focused on clinical trials. In an HIV clinical trial, 91% of patients surveyed on potential methods to decrease consent form length agreed with using appendices that would contain supplemental information, and 65% agreed that reading the appendices should not be required for IC (Corneli et al., 2017). The Clinical Trials Transformation Initiative recommended the use of a tiered consent form to help improve the IC process. The idea of using tiers is a similar approach to using appendices in that the first tier would provide the information critical to IC, and subsequent tiers would hold supplemental information (Lentz, Kennett, Perlmutter, & Forrest, 2016). The published literature also supports the use of videos in the consenting process. A study investigating consent for genomic data sharing, found that a video supplementing a one page consent form improved understanding in members of the general population (Riggs et al., 2019). A video consent, administered via tablet, that was designed for a study assessing diabetes risk was found to be significantly associated with greater comprehension scores as compared to a standard paper consent form (Lindsley, 2019). A study evaluating the consent process in Australia interviewed stakeholders, including 14 research participants, and concluded that is important to have multiple ways to communicate study information (such as consent forms, videos, and in-person conversations) as not every research participant will have the same needs (McWhirter & Eckstein, 2018). We recommend that the permissible formats of consent forms by REB and other governing bodies be updated to include the use of appendices, alternate information mediums (such as videos),

and allow online versions of consent forms. Utilizing online consent forms may be attainable already for some studies as most institutional review boards (IRB) chairs have indicated that they are open to accepting electronic signatures from research participants (Kane & Gallo, 2017). Research groups could also consider the use of patient decision aids, which provide accurate information regarding a health care decision and assist patients in making an informed choice based on their values and preferences. Decision aids have widely been shown increase to patient involvement in decision making, improve patient knowledge scores and accuracy of risk perceptions, and decrease decisional conflict (Stacey et al., 2017). Adam et al. (2018) found patients undergoing GWS who were randomized to receive genetic counseling or DECIDE, an online decision aid, had equivalent increases in knowledge. Our study aimed to gather the preferences of parents and HCP on the content of consent forms for GWS, and did not assess knowledge outcomes, decision making, or decisional conflict/regret.

In all forms of communication, it is important to use language appropriate for the patient population. However, the language in consent forms is largely determined by HCP and REB, or IRB, members. The REB/IRB is an important stakeholder in the content and complexity of consent forms. IRB chairs have reported that they are concerned with patient understanding during the IC process, but in practice most IRB rarely monitor studies for patient understanding, and do not regularly assess if consent forms are meeting the recommended reading level (Kane & Gallo, 2017). The perspectives of IRB members, researchers, and research participants can differ with respect to which aspects of consent forms are important. IRB members, on average, marked a larger portion of a consent form for biobanking as important (72%), compared to researchers and research participants only marking 53% and 40% of the consent form as important, respectively (Beskow, Friedman, Hardy, Lin, & Weinfurt, 2010). The contrasting views among stakeholders, in addition to the preference of IRB members for longer and more detailed consent forms, may make simplifying or modifying consent forms in the future challenging. One study recommended that researchers and IRB professionals collaborate to create templates for research materials after interviews with 31 IRB professionals indicated that they are not comfortable their own understanding of genomics, or independently creating research guidelines for the return of GWS results (Dressler et al., 2012). The GHP in the current study were able to recognize text written by the REB and felt that the REB required text was often unclear, lengthy, or not appropriate for GWS consent forms. For example, one section of REB required text stated that data produced by the study belonged to the corresponding study participants, which in the context of GWS, does not provide a clear answer to patients or HCP as to whether or not studies are expected to provide raw sequencing data to patients. This ambiguity was raised by both PP and GHP in their respective focus groups, with some PP expressing interest in having this data. All stakeholders should be involved in the design and implementation of research studies. This should include parent and patient representation on REB/IRB committees in order to provide input on content of consent forms and other study materials. Consent forms have been used to communicate study information as well as being a way to document IC, and parent and patient representatives should help determine what qualifies as appropriate and understandable language.

The study reports the novel finding that PP used the consent form as a resource throughout and after completion of the study. Parental empowerment came from information in the consent form enabling research and GWS decision-making independent of the pre-test counseling session. Importantly, the parents who participated in the focus groups received the consent forms before the pre-test counseling appointment as was the standard procedure for the CAUSES Study. Information on connecting with other families was not included in the consent form. The peer support requested by PP in the current study supports research that has investigated the perspectives of parents whose children have received GWS or have a rare disease. Interviews with parents whose children underwent GWS found that many parents wanted to connect with families who had a child with the same or a similar diagnosis to their own child (Rosell et al., 2016). Peer networks for parents of children with rare diseases were important for emotional support, education and accessing resources. Parents often connect with families through online platforms and social media, looking for rare disease-specific groups, or searching by syndrome name if their child has a diagnosis (Baumbusch, Mayer, & Sloan-Yip, 2018; Inglese et al., 2019; Krabbenborg et al., 2016). It is important for parents to be connected to a disease-specific peer group when possible, as a diagnosis from GWS may have the unintended effect of isolating parents from their undiagnosed rare disease support group (Baumbusch et al., 2018). As disease-specific information cannot be provided in the consent form it is critical that resources be discussed during the results session. An important role of genetic counselors and clinical geneticists is to identify support groups, including non-traditional support groups on social media before the results session. Due to the rarity of the genetic diagnoses that can be obtained through GWS it may not be possible to refer patients to diagnosis-specific support groups or communities at the time of the diagnosis. Providing patients with resources such as MyGene2 (www.mygene2.org) and Unique (www.rarechromo.org) would enable them to connect with families by diagnosis in the future. Parents of children who received a variant of uncertain significance (VUS) also perceived benefits in support from peers with a shared experience and wanted to be connected with families whose child received the same category of result (Li et al., 2019). For patients who did not receive a

diagnosis from GWS, or for whom a disease-specific support group is not available, providing families with more general resources, such as the Peer 2 Peer Resource Network from the Rare Disease Foundation (www.rarediseasefoundation.org) enables them to connect online.

5.1 | Study limitations

Our study could have an ascertainment bias as the parents and genetic HCP who agreed to participate in this study may be more likely to be interested in the consenting process for GWS. As such, it is possible that additional themes could emerge from this study population with additional focus groups. People who participate in genomics research have a significantly higher information need and greater information seeking behavior (Dijkstra et al., 2010). This may explain the high value placed on detailed information by our PP. The participants in both focus groups consisted primarily of females and of people with European ancestry, consistent with the limitations seen in other genomics research (both GWS and non-GWS studies) (Hensley Alford et al., 2011; Hindorff et al., 2017).

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CONFLICT OF INTEREST

Authors Emma C. Hitchcock and Alison M. Elliott report no conflict of interest.

AUTHOR CONTRIBUTIONS

Emma Hitchcock contributed to the research design, data acquisition and analysis, wrote the initial manuscript, and contributed to revision of the manuscript. Dr. Alison Elliott contributed to the research design, data acquisition and analysis, as well as critical revision of the manuscript. Both authors approved the final version to be published and agreed to be accountable for all aspects of this research.

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