

Enhancing Genomic Laboratory Reports from the Patients' View: A Qualitative Analysis

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The purpose of this study was to develop a family genomic laboratory report designed to communicate genome sequencing results to parents of children who were participating in a whole genome sequencing clinical research study. Semi-structured interviews were conducted with parents of children who participated in a whole genome sequencing clinical research study to address the elements, language and format of a sample family-directed genome laboratory report. The qualitative interviews were followed by two focus groups aimed at evaluating example presentations of information about prognosis and next steps related to the whole genome sequencing result. Three themes emerged from the qualitative data: (i) Parents described a continual search for valid information and resources regarding their child's condition, a need that prior reports did not meet for parents; (ii) Parents believed that the Family Report would help facilitate communication with physicians and family members; and (iii) Parents identified specific items they appreciated in a genomics Family Report: simplicity of language, logical flow, visual appeal, information on what to expect in the future and recommended next steps. Parents affirmed their desire for a family genomic results report designed for their use and reference. They articulated the need for clear, easy to understand language that provided information with temporal detail and specific recommendations regarding relevant findings consistent with that available to clinicians. © 2015 The Authors. *American Journal of Medical Genetics Part A* Published by Wiley Periodicals, Inc.

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INTRODUCTION

Genetic disorders as a group are common, and affect patients and families throughout their lives. Yet, patients and their providers struggle to have ready access to the information

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needed for appropriate management and coordination of care. Patients' knowledge contributes to successful chronic disease management because, '...by living with and learning to manage a long term illness many people develop a high degree of expertise and wisdom'[Wilson, 1999]. Such knowledge is even more critical in rare diseases where the disorders are not only chronic, but typically lack treatment guidelines.

Lack of rapid access to accurate genetic information is a significant barrier to the pursuit of appropriate care for patients with genetic disease [Levy et al., 2008]. The genetic-laboratory report could be used as a tool; however, its current purpose is solely to transmit the results of laboratory tests to providers. As a result,

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genetic test reports contain information about changes in DNA structure, with the expectation that the provider will have the required knowledge to interpret the results. This technical language is a challenge for providers outside genetics, let alone patient families, leading to errors that can negatively affect patient care [Emery and Rimoïn, 1990; Wilson, 1999; Rare Disease Foundation]. Laboratory reports have the potential to provide information at the point of care that leads to improved patient outcomes [Hammond and Flinner, 1997; Laposata et al., 2004]. Results from studies of provider reactions to genetic reports led to a new format for genetic test reports [Lubin et al., 2009; Scheuner et al., 2012]. Testing of this report showed significantly higher satisfaction, ease of use and efficiency for the formatted genetic test report compared to standard reporting [Scheuner et al., 2013]. An opinion article by Haga et al outlined many of the challenges in developing patient-friendly genomic test reports and suggested four potential options to reformat laboratory reports aimed for patient use.

Our study approach most closely resembles their option to design “a completely revised patient friendly report” [Haga et al., 2014]. Similar to their conclusion, we suggest that a new patient-centered genomic Family Report could improve shared decision-making between the patient/family and providers, as well as improve care and outcomes. This new Family Report should convey genetic information to patients—and their doctors—in a way that enhances understanding, promotes engagement, and provides the basis for meaningful decision-making. We present the results of a study undertaken to learn the type of information deemed essential by the parents of children involved in a clinical research study of whole genome sequencing (WGS).

METHODS

The Institutional Review Board of the Geisinger Health System reviewed and approved this study’s protocol. Semi-structured interviews were conducted with parents of children who participated in a whole genome sequencing clinical research study to address the elements, language and format of a sample family-directed genome laboratory report. The qualitative interviews were followed by two focus groups aimed at evaluating example presentations of information about prognosis and next steps related to the whole genome sequencing result. Semi-structured interviews, followed by a second round of focus groups to conduct a descriptive qualitative analysis were chosen because little previous work has been published to inform development of a patient-facing genome results report.

Study population

The participants for this study were recruited from participants in a study examining the use of WGS for children with intellectual disability, with or without other medical conditions, for whom there was no causal diagnosis despite extensive prior evaluation. The participants in the study were the parents of those children. The WGS project required participation of both parents; therefore, enrollment in this study was offered to both fathers and mothers. There were 42 couples available for recruitment. Thematic saturation was reached after the eighth individual participant. Researchers conducted another inter-

view (#9) to confirm that no new relevant data emerged in relationship to the research question [Scheuner et al., 2012].

Genome Family Report Preparation

A sample Family Report was developed for the qualitative interviews and was based, in part, on a pre-existing provider report that met published recommendations and submitted as part of the SimulConsult/Geisinger response to the Boston Children’s Hospital CLARITY competition which involved creating standardized methods for analyzing, interpreting, reporting and, using genomic information in a clinical setting [Zallen, 1997; Lubin et al., 2009; Scheuner et al., 2012; Scheuner et al., 2013; Brownstein et al., 2014]. Modifications were made to simplify readability based on health literacy principles and previously published studies on communicating genetic risk information to patients [McGee, 2010; Lautenbach et al., 2013].

During the interview phase, parents reported that more prognostic and condition-specific information regarding next steps was needed; however, they were not able to articulate how this could be accomplished. Initially, we designed the “Next Steps” section of the Family Report to facilitate a conversation between the family and provider about the appropriate next steps for the individual child in the context of the results. Rather than explicitly listing “next steps” for the child, we offered a list of topics to address, and specific questions for the family to ask their physician. In the original example report this section was designed to be applicable to any genetic finding, so even though our example report pertains to a diagnosis of Salih myopathy, there are no items in this section that reference the specific condition: (see Supplementary materials, Family Report: Next Steps section)

Parents said that wanted more information than what was provided. Specifically, they wanted to know about “what to expect in the future” and any action steps that were appropriate for a child with this type of genetic testing result. We decided to conduct focus group discussions to study how best to convey this complex disease-specific information, we designed four example Concept Sheets. The sheets were developed with the support of the SimulConsult Diagnostic Decision Support software to include frequency and time course of findings in diseases. This information was incorporated into four different visual representations of the prognosis and care information, labelled as concept sheets 1–4 (see Supplemental Information, Concept Sheets).

In addition, a few participants commented that they would find it easier to evaluate a report for an example patient whose symptoms were more similar to their own children’s symptoms. Our participants were parents of children with intellectual disability, but the example report described a child with a neuromuscular disorder. Since the discussions would specifically address how to best present information about symptoms, we decided to use a different genetic condition as the topic for the Concept Sheets—Mowat–Wilson syndrome—so that intellectual disability would be a major feature described.

Procedures for Qualitative Data Collection

All interviews were conducted in person by team members trained to standardize the interview process. The Family Report

was provided to participants approximately three days prior to the interviews. Using an interview guide, eight individual interviews and one couples interview were completed. The sessions lasted from 35 to 65 min (average 50 min.) Parent participants were asked to comment on areas of focus including potential improvements to the Family Report, success in communication, and unmet needs related to the communication of genetic information.

Following the request by parents for more specific prognostic information, we conducted two focus groups with a total of five participants. Of those, three of the focus group participants had also participated in the semi-structured interviews. The four concept sheets were presented in both focus groups. Participants were asked to provide feedback on whether they found the information useful and to recommend the best way to represent this complex prognostic information.

Audio recordings were made during the interviews and the focus-group discussions. These recordings were transcribed and analyzed using Atlas.ti 7 software. A coding dictionary of “meaning units” (ideas that seem to fit together) was created, data were coded and no data were double-coded. Based on the coding of two interview transcripts, inter-rater reliability was very good ($k=0.85$). The findings were reported using an interpretative phenomenological analysis, where the individual’s perceptions about an object or event are reported and analyzed in this case summarized as: “What are the factors of the genetics report that would hinder or improve the patient’s experience of their child’s illness?” [Packer and Addison, 1989].

RESULTS

Three themes emerged from the qualitative data obtained in the semi-structured interviews and the focus group discussions: (i) Parents described a continual search for valid information and resources regarding their child’s condition, a need that prior reports did not meet for parents; (ii) Parents believed that the Family Report would help facilitate communication with physicians and family members; and (iii) Parents identified specific items they appreciated in a genomics Family Report: simplicity of language, logical flow, visual appeal, information on what to expect in the future and recommended next steps.

Theme One: Parents Described a Continual Search for Valid Information and Resources Regarding Their Child’s Condition, a Need That Prior Reports Didn’t Meet

The parents involved in this study had been through multiple diagnostic testing rounds that ended without a diagnosis. Some said they were “*numb*” from searching for a diagnosis for over a decade of frequent tests.

“...over the past 16 years. . .at first it was a little nerve-racking. I know I was scared. . .when they first started doing testing, I dreaded it, I was like on the phone, did you get the results back, yet? And now, it’s kind of like, okay well when they come.” Participant #1403

When asked about previous experiences with actual laboratory reports, some parent participants indicated that they had received a laboratory report of some type but it wasn’t very helpful.

“...a printout one time. It was just a copy of like the X chromosome things . . .and like you could see the differences in some of it but that was it. There was no explanation.” #1407

The Geisinger patient portal was also mentioned, but it “*just contained a copy of the results with no explanation.*” Three families could not recall receiving copies of any laboratory reports at all. Most parents described that they had received a written summary letter describing genetic testing results and that previous results were most helpful when discussed in-person with a provider.

Parents expressed resignation at the lack of a diagnosis, and viewed uncertainty “*like the norm.*” Although parents said their child’s condition “*is what it is,*” they described looking for additional resources in hopes of finding some relevant information or a diagnosis that seemed to match their child’s symptoms. They searched resources broadly, including a range of popular magazines to medical journals. As technology progressed, the majority searched for information online and on various condition-specific websites, including (i) the National Rett Syndrome Association; (ii) Angelman Syndrome website; (iii) WebMD; (iv) Mayo Clinic website; (v) and the National Library of Medicine. One parent called the internet “*a blessing and a curse.*” At least one found online searches “*scaring herself*” more than helping. Others made searching an ongoing project, as one mother said:

“*At one point I had a file of things printed and highlighted. Yes, this was [child’s name], no this wasn’t (. . .). Then I would re-read it. Did I still agree with what I had highlighted? Yes this was (...). It was like my little research project.*” #1401

Determining the reliability of information was a key concern. When parents doubted what they found on the Internet, they reported discussing the information with a family member who was familiar with medicine/genetics or their child’s medical provider.

Theme Two: Parents Believed That Building a Family Report Would Help Facilitate Communication with Physicians and Family Members

Parents believed the Family Report would be important to review in conjunction with a provider to discuss specific expectations and management needed for the condition. Without prompting, parents described how they would share the Family Report with family, friends, and physicians, because it was a vehicle for communication about their child’s condition. One mother said,

“...where it would be nice to have it all in a report for us . . . and if we wanted to take it to our family doctor, we could take it wherever or even family members and have them look over it and have them understand it, that would be great.” #1404

Parents believed that they could use the Family Report to look at the information later, after the physician appointment, because parents will not remember everything talked about in the visit because “*you’re thinking a thousand other things in your head.*”

Another participant was pleased that the sample report included a section about issues to discuss with your doctor:

“...cause sometimes you can't always think maybe of what you want to say. . . when you get in to the appointment you forget, so it's kind of just take the paper right with you and just ask the questions.” #1407

Theme Three: Parents Identified Specific Items They Appreciated in a Genomics Family Report: Simplicity of Language, Logical Flow, Visual Appeal, Information on What to Expect in the Future and Recommended Next Steps

The most frequent comment about the Family Report was that it was easy to understand. For example, it was *“straightforward, used everyday language”* and *“not words that I have to look up.”* One parent said:

“It didn't get very technical, 'cause like I said earlier, if it gets too technical. Yep, it's going right over my head, I'm not going to understand it, but this seemed to be a good level that I could understand.” #1405

Parents appreciated the linear organization of the report and noted the visual appeal.

Two visual aids were provided in the report to help illustrate inheritance patterns, and all parents believed the illustrations were easy to understand. However, parents were split between a cartoon-like image being *“too cute”* for a medical report, and another silhouette design being too impersonal and confusing.

“I look at this and I kind of feel like this is something you might use on a 6th grade science class, and that would be effective for that age group . . . but not for adults ”#1408

Two new examples of inheritance patterns were presented to the focus groups to solicit feedback. Participants affirmed that inheritance pattern diagrams were an important illustrative element to show family inheritance, but must be explained in the text. One parent cautioned to label the diagrams carefully, stating:

“It's not about assigning blame, but you may have people who get to this point and go, oh it's my fault, my gene did it and then you're going to have guilt. . .” #1401

Other parents considered it important to know which side of the family the variant is associated with because siblings or offspring may want to be informed.

Parents preferred using numbers, followed by percentages, such as *“1 chance in 4, or 25%”* rather than displaying only one or the other. Similarly, parents preferred using the both the technical word and the definition, such as myopathy (muscle disease). Most parents appreciated the glossary within the Family Report.

Word choices mattered. Participants preferred *“cause or probable cause”* to pathogenic. One mother said that *“pathogenic sounds scarier than likely to be the cause”* and another admitted that he simply did not know what the term meant. In the Family Report, we used two different words *“variant”* and *“mutation”* for the actual change in the genes. Participants wanted consistency and preferred the term *“variant”* because *“mutation indicates there's something wrong there, something not normal.”*

All parents wanted secondary findings to be included in the same report as the primary findings. In our Family Report, the secondary finding was a *BRCA1* mutation. Parents responded that knowing additional genomic results would help them better plan for the future and seek appropriate help. Even though the additional findings section made the report twice as long, all parents wanted all the information at the same time, but separated from the primary results. One mother said, *“I don't think it can be too long when it's somebody's kid.”* Another commented that *“After I got grounded and was able to sit down and read through this and I would be thankful because it's very thorough.”*

In the section on *“Next Steps,”* parents desired more information about prognosis, verbalized as *“what to expect in the future”* even if the condition was *“going to get worse.”* They wanted more information about what to do next in clearly delineated action steps for their child. A parent stated, *“I'd probably say, so what are we looking at in the future with this? What does this involve?”* Parents wanted a step-by-step final page that provided anticipatory guidance, including seeking speech or other therapies, specialists, or even house modifications. Parents commented:

“Because I think that's one of my biggest questions looking back over the years, is okay what do I do with this information you just gave me?” #1408

“..I would want to know what I should do next, you know, regarding him and his health and you know there is someone who I should see specifically, people in the world of medicine, you guys know that we may not. . .” #1409

Parents appreciated the section on resources and support groups. They wanted to know whom to contact or numbers to call, so they didn't have to *“go home and figure out how to find that information”* and so *“you're not googling for 5 hours.”*

Focus Group Component

The symptoms of genetic diseases typically unfold over time and for most genetic diseases, many, if not most, of the signs and symptoms that are characteristic of a disease are only experienced by a portion of those with the diagnosis. Thus, the information about what to expect is complex. Participants were presented with four different concepts, three concepts consisted of tables that presented prognosis information in different ways and one was a text-based sheet about appropriate next steps. Results of the concept sheet evaluation are summarized in Table I. Parents wanted a combination of the table-based and text-based concepts. Although they did not know some of the medical words used, the comprehensive table-based Concept 4 was the most preferred of the three table-based concepts because *“it's very specific”* and they would *“like to see how many of those symptoms that my child would have.”*

Parents wanted the information from the text-based example (Concept 3), ranking it as their first or second preference because of the way it *“gives recommendations or a plan to follow.”*

“I do like how it's broken down into the different categories so it makes it kind of easy even if you just want to scan it really quick to find out what you're looking for.” FG2P1

TABLE I. Focus Group Results: Prognosis Concept Sheets

Evaluation concept sheets	Appreciated	Not Appreciated	Suggestions
Concept 1	<ul style="list-style-type: none"> —Gives idea of what to look for in the future —Timeframes very helpful —Would use as baseline for reference —Monitoring recommendations —Everything on it is necessary —Can use with provider for discussion 	<ul style="list-style-type: none"> Classifications: Few, Some, Most 	<ul style="list-style-type: none"> Use percentages
Concept 2	<ul style="list-style-type: none"> The findings are ordered by likely possibility 	<ul style="list-style-type: none"> —Difficult to read —Must read every word to understand —Don't like anything about concept 	<ul style="list-style-type: none"> Perhaps separate into columns to make easier to read
Concept 3	<ul style="list-style-type: none"> —Clearly divided into sections —Can scan quickly —Recommended specialists to see —Answers concern of where to go next Provides accepted listing of special needs 	<ul style="list-style-type: none"> Not color coded Too much black text on white background 	<ul style="list-style-type: none"> Provide actual names of specialists in the healthcare system
Concept 4	<ul style="list-style-type: none"> —Very detailed and specific —More information is better —All the findings available not just the "important" ones —Allows for discussion of behavioral and physical findings 	<ul style="list-style-type: none"> medical terminology 	<ul style="list-style-type: none"> —Need to define all medical words —Offer functionality to check off symptoms that match

Parents also appreciated the specific guidance offered in the sections related to monitoring, support services, and additional medical specialists.

"...this last one here helps you to know... where you can get more help... it is helpful because it is giving you different ways you can go... for the support services, so that you know that you're not alone...." FG1P1

Parents suggested that color could improve the Concept 3 presentation because otherwise it could get lost as "white noise"; *"if you set this down with other white papers around it, what's going to happen to it? You're going to lose it."*

None of the parents liked the categories "few," "some," and "most;" used in two of the table-based concepts stating: *"When I look at this I think I want to see percentages. When I see like at one month "most" [kids are affected], I want to know like is it like 75%, like what is "most," is it...?"* and *"My question too, what does "few" mean and what does "some" mean? What [is] the difference between those?"*

All parents wanted a timeline that spanned birth to 15 years.

"...but I know to get information like this when your child is 1 year old, it would help to know what... has happened with others... of what to look at in the future." FG2P1

"To kind of look at the future, like what you might expect or what you kind of keep your eyes open for or what you might be looking for. I want to know what I'm going to do next if this [finding] occurs" FG2P4

When asked about how best to present this complex information, parents in one focus group suggested solutions involving a website or thumb drive to provide the desired information because *"you could download it"* or *"you can print out the information you want."* Parents also thought this solution would allow them to use the information on their phone or iPad so *"you have it with you all the time that if you go to the doctor you can just pop it up."*

Furthermore, parents discussed at length how they would use the information to facilitate communication with doctors, family

members, other caregivers, and schools. Regardless of how the information was presented, the participants wanted to get the results back as soon as they were available without delay. One woman described, *"... if I knew you did a test and the test results are back today, I want to know today."* One set of parents summarized, *"[The report] gives you the basics that you would need to know"* and *"it gives us the inspiration we need."*

DISCUSSION

Several recent initiatives support increased patient involvement in defining value and quality in the healthcare they receive. The Patient Centered Outcomes Research Institute, formulated as part of the Affordable Care Act, was established to define patient centered care using methods of direct patient engagement in the research process. In this study the outcome desired was identification of the elements deemed critical in a genomic result report. The patients were individuals who had been through multiple episodes of genetic testing for their children. Their previous experience of receiving genetic test results often did not include receiving a copy of the actual laboratory report. Those who had received reports described their difficulty understanding them. The presentation of the sample Family Report that combined the genomic test results with comprehensive information about the condition was enthusiastically endorsed.

Participants appreciated the simple, easy to understand flow of language as well as the visual layout and sections of the Family Report. They articulated that diagrams help, color is engaging, and multiple pages are fine when the information is categorized into discrete sections. Specifically participants highlighted the glossary, the use of medical terms and "layman's" explanations as extremely successful in this sample Family Report.

The original Next Steps section was found to be insufficient, though the suggested questions were helpful. At the end of the

interview when asked, “*What is missing or what would one change about this Family Report?*”, most responded with the request for more specific information regarding what to expect in the future as well as the specific next steps to take. The parents wanted to know what to do with the test result, and wanted the Family Report to be action oriented. One parent summarized that providers know things that parents don’t know about their child relative to this specific diagnosis, or where to go to get expert information. She urged that medical professionals share that information with parents.

Parents appreciated the usefulness of a Family Report written for them, envisioning use of the report at an Individualized Education Program planning meeting, sharing with family members to help to understand their child’s characteristics, and using the list of findings to review with their provider to identify potential future issues that will need anticipatory care. Finally, several participants commented that they would be relieved to have a written report that they could go back to over time, recognizing that at the initial moment when information is shared, it may be too much to absorb. The Family Report allows recipients to review and re-review the report at their leisure.

Limitations

Many of the study participants have been searching for the diagnostic explanation for their child’s condition for many years. The desires articulated by these individuals may not be the same as individuals who are new to diagnostic testing. It is possible that the participants were most representative of those who want to know more.

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SUPPORTING INFORMATION

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