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Enhancing Genomic Laboratory Reports: A Qualitative Analysis of Provider Review

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This study reports on the responses of physicians who reviewed provider and patient versions of a genomic laboratory report designed to communicate results of whole genome sequencing. Semi-structured interviews addressed concept communication, elements, and format of example genome reports. Analysis of the coded transcripts resulted in recognition of three constructs around communication of genome sequencing results: (1) Providers agreed that whole genomic sequencing results are complex and they welcomed a report that provided supportive interpretation information to accompany sequencing results; (2) Providers strongly endorsed a report that included active clinical guidance, such as reference to practice guidelines, if available; and (3) Providers valued the genomic report as a resource that would serve as the basis to facilitate communication of genome sequencing results with their patients and families. Providers valued both versions of the report, though they affirmed the need for a provider-oriented report. Critical elements of the report included clear language to explain the result, as well as consolidated yet comprehensive prognostic information with clear guidance over time for the clinical care of the patient. Most importantly, it appears a report with this design has the potential not only to return results but also serves as a communication tool to help providers and patients discuss and coordinate care over time. © 2016 Wiley Periodicals, Inc.

Key words: genomics; laboratory reports; provider communication; patient communication

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INTRODUCTION

Most rare genetic disorders are chronic and affect patients and their families throughout their lives. Once diagnosed, the challenge for patients, families and their providers is having ready access to the information necessary for appropriate management and coordination of care. Health-care providers who are not genetic professionals often are uncomfortable interpreting genetic test results or explaining genetic disease. [Bonter et al., 2011; McLaughlin et al., 2014] This issue will become more problematic as exome and genome analysis assume a larger role in healthcare and the information from all of an individual's genes (genomic information) is used to diagnose and manage medical conditions. In the past, the purpose of most laboratory reports was simply to transmit results of laboratory tests to providers. As a result, genomic test reports contain information about genetic changes that require significant content knowledge on the part of the provider to appropriately interpret the results. This technical language is a challenge for providers outside genetics; leading to errors that can adversely affect patient care [Lebo and Grody, 2007; Bonter et al., 2011; Haga et al., 2014; Vassy et al., 2015].

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Recent studies have shown the potential for laboratory reports, when designed differently, to provide information for clinical decision making that can lead to improved patient outcomes. These studies utilized extensive input from genetics and non-genetics providers to create improved laboratory report templates. Significantly higher satisfaction, as well as ease of use and efficiency for the formatted genetic test report compared to standard reporting were noted in initial testing of the draft template. In fact, the greatest benefit of a differently designed template was seen for providers least familiar with genetic laboratory reports. Appropriately designed genomic laboratory reports, therefore, have the potential to provide information at the point of care and lead in turn to improved patient outcomes [Lubin et al., 2009; Scheuner et al., 2012, 2013; Dorschner et al., 2014; Vassy et al., 2015].

However, it has not been shown that this leads to improved communication between providers and patients, which is what is needed if genomic results are to be used to improve care over the long-term. We hypothesize that genetic laboratory reports designed with input from patients as well as providers can be used to promote and enhance communication and decision making between patients and providers. Previously, we reported on the development of a family genome results report that used patient and parent responses to inform design and content. [Stuckey et al., 2015] Here, we report on provider response to a genomic report designed to communicate the results of whole genome sequencing.

METHODS

We conducted semi-structured interviews with six healthcare providers to review elements, language, and structure of a fictional genome results report. In addition to the provider version of the report, participants were given the opportunity to review the genomic report designed for patients. A single interviewer met with all participants and reviewed each section of the report on a section by section basis to facilitate comments throughout. After reviewing the provider report and eliciting all focused and overall feedback and impressions, providers were given the opportunity to review and comment on the previously developed patient genome report [Stuckey et al., 2015] and were asked whether a single report could be sufficient to meet both the provider and family needs.

Study Population

The participants included a small convenience sample of physicians at Geisinger. None of the provider participants interviewed had formal training in genetics.

Whole Genome Sequencing Results Report

The genomic report developed for use in this study derived from the clinical genome report designed and submitted by the joint Geisinger and SimulConsult® team in response to the Boston Children's Hospital 2013 CLARITY Challenge competition [Brownstein et al., 2014]. The original Clarity report included: a) prominent placement of the primary genomic findings; b) hyperlinks to OMIM, GeneReviews, and other resources for genomic findings; c) hyperlinks to patient and family support resources; and d) technical specifications. Modifications to the original report consisted of: a) clear concise non-genetics language whenever possible; b) summary of clinical findings; c) direct guidance for clinical implications of the genetic result, prognosis (such as published guidelines when available); d) temporal disease prognostic information in table format for additional clinical guidance; and e) research section devoted to variants of research interest. The team considered suggestions contributed by reviewers of the patient genome report [Stuckey et al., 2015] to inform revisions predicted to improve provider understanding and facilitate provider-patient communication about test results.

The report contained 15 sections (Table I) organized in standard laboratory format with primary results prominently located at the top of the report. Key technical information such as laboratory specifications, analysis information, and variant classification was located at the end of the report. The report in the current study presented a fictional scenario of a 10-year-old boy found to have a *TTN* mutation as the primary finding explaining the child's clinical presentation. It also included a secondary/incidental finding of a *BRCA1* mutation. The report contained fictional variants reported as primary and incidental findings generated for the purpose of provider review.

The genome report was sent via email to providers at least two days prior to the scheduled interview. A printed copy of the report was provided for the interview visit. (Report can be found in the supplementary materials). Providers were informed that an actual report would contain active hyperlinks to online resources specific to the genomic finding, whether primary or incidental results, and the report would be accessible in the electronic health record (EHR) of their patient at the time of the patient visit. They were also told that patients could access a patient version of the report through the EHR patient portal.

All interviews were audio recorded, transcribed, reviewed and coded within each of the report sections. The coded responses were analyzed in the framework of qualitative description, in which the participant's perceptions about an object or event are reported [Sandelowski, 2001]. The central issue connecting all

TABLE I. Genome Report Sections

Patient demographic and provider information
Primary finding
Clinical rationale
Secondary (incidental) findings
Clinical rationale
Confirmatory testing
Clinician resources
Patient resources
Research and clinical trials
SimulConsult® patient clinical summary
Prognosis table©
Next steps: Care management
Inheritance and family implications
Comprehensive gene variants list
Technical documentation of sequencing methods

phases of this study was: "What factors of the genomic report hinder or improve the provider's experience in understanding and communicating genomic results with their patients?" [Packer and Addison, 1989]. Although 10 providers were recruited for the study, after five providers, the same data began to repeat (saturation) [Morse, 1995]. We continued with another interview for confirmation that no new data emerged. Two study staff reviewed all transcripts with the audio files for accuracy and completeness (JLW, AKR). Transcript text was coded in relation to the sections of the report by one reviewer (JLW). Three reviewers (JLW, HS, AKR) reviewed the section texts and developed the overarching constructs of codes resulting from providers' response to each section.

This study protocol was reviewed and approved by the Geisinger Health System Institutional Review Board.

RESULTS

Six physicians participated in semi-structured interviews. Three physicians indicated that they reviewed the emailed genome report in advance of the interview. Areas of expertise of the participants included pediatric subspecialties and internal medicine. Over half the physicians were male, and two thirds had more than 10 years practice experience as shown in Table II.

Analysis of the coded transcripts resulted in the recognition of three constructs around communication of genome sequencing results: (1) Providers agreed that whole genomic sequencing results are complex and they welcomed a report that provided supportive interpretation information to accompany sequencing results; (2) Providers strongly endorsed a report that included active clinical guidance, such as reference to practice guidelines, if available; and (3) Providers valued the genomic report as a resource that would serve as the basis to facilitate communication of genome sequencing results with their patients and families.

Whole Genome Sequencing Results Are Complex and Supportive Interpretation Information Is Critical

In general, providers recognized the format of the genome report as similar to genetic laboratory reports they had received in the past. They noted that the first page structure, which included the primary genome result at the top of the page, followed by the laboratory interpretation, was familiar. However, providers also commented that they hadn't considered the idea that genome sequencing results would include the need to report incidental findings, and supported the concept that incidental findings should appear proximate to the primary result so that such results would not be missed.

TABLE II. Provider Characteristics

Specialty	Male/Female	Practice experience
Internal medicine/pediatrics	Female	<10 years
Internal medicine/pediatrics	Male	<10 years
Pediatric cardiologist/intensivist (researcher/non-clinical)	Female	>10 years
Pediatric specialist: Neurodevelopmental	Male	>10 years
Internal medicine	Male	>10 years
Pediatric specialist: Gastrointestinal	Male	>10 years

All providers reported they were not familiar with the gene or the condition reported as the primary genomic result in the report. Five providers commented that the Clinical Rationale section of the report, which provided an explanation of gene function and a brief summary of clinical information, improved their understanding of the gene result, as in comments by two providers:

> *". . . when you first get it if you're with the patient . . . you would want to know what they have and the significance of it. . . where it tells you quickly what's in store."*

> *"I like the level of detail that it gives, not excessive, .. but enough."*

They appreciated the inclusion of contextual links, positioned with the result information, to provider-related condition-specific resources. Two providers appreciated that links included went directly to the relevant material for the diagnosis in OMIM and GeneReviews as evidenced by the response of one:

> *"I can look at the report and ... click a link. I actually have a better idea when I bring the patient in to know what to tell them and then offer resources."*

Three providers stated overall that this sample genomic report was better than any other genetic test report they had received in the past. The design and content was described as "*perfect*", "*simple*", and "*clear*". *"I think this looks great, and I think it is much more helpful than what I currently receive."*

Providers also gave feedback regarding the presentation and usability of the **Technical** section of the report. Overall, they reported they were uncertain that they would use the sections reporting laboratory technical information, definitions of pathogenicity, and variants for future research, but recognized the importance of including the laboratory technical information documenting the specifics of the sequencing and analysis for future reference. Two providers related their past experience with the changes in microarray technology and the need to understand what each technology detected and what could have been missed as examples of why this information is important to include.

One provider recommended that the report include definitions of variant classification clarifying how gene variants get labelled as "pathogenic" or "likely pathogenic" because providers may want to know what criteria are used and how they are applied to reach these classifications.

> *"'This is likely pathogenic' to you means something very specific, but to the clinician that hasn't been trained, they don't know that."*

The section of report which listed variants with potential research investigation relative to the primary indication for sequencing was problematic for providers. This section was developed to address a circumstance in which no pathogenic or likely pathogenic variants were found to explain the referral indication for sequencing. This list of variants could be used to direct future re-analysis or focus opportunities for research. One participant expressed the general feeling all providers.

> *"I have no clue of what it means or what it does, so I don't know that I'm going to go there. . . ..But it is good information that should be in the record. I think it must be there but it's just in the background."*

Providers acknowledged the importance of variant information to genetics providers, and that the information might guide re-analysis over time. However, they were concerned about what they were to do (action steps) with the list of variants. One provider noted:

> *"So as a clinician I'm going to be saying, here's this list, now let's see, I get a list of* [variants of] *unknown significance, . . .well what am I going to do with this?"*

Several providers indicated concern about their responsibility to stay on top of the list of gene variants as new information became known. They asked how a clinician will know to look back at the gene variant list and recognize that there is a gene variant that now applies to their patient.

> *"So . . .as things start to become known, is there a way to reconnect . . .now we know what DLRA4 gene is and it may be related to this [child's diagnosis]. So is there a way that this is updated or the clinician will know—or is it just . . .you have to hope. As a clinician this is going to be frustrating ... how do we make sure that there's that ongoing communication and know to . . .at least think about it again in the future."*

Concern about potential liability for not evaluating the list and warning the patient of possible health implications was expressed.

> "*I have reviewed this report, so I have taken full charge of this. Now 2 years down the road, the gene is associated with some horrible disease, and I don't think of this all the time... I can imagine a lawyer in a court of law saying doc you had the list of variants. . .Didn't you see it?*"

To address this concern, several providers suggested the report include information on what to do with the research variant list. Suggestions included electronic options for searching the variants in order to identify new associations for their patient and that a disclaimer should be added to document the need for re-analysis of the variant findings. Two providers also requested that date stamps be prominently visible with regard to all information in the report.

Reports That Include Active Clinical Guidance, Such as Reference to Practice Guidelines Is Highly Valued by Providers

Providers indicated that they envisioned themselves accessing this report in the patient's EHR during a clinic visit. Most admitted that it was unlikely that they would have reviewed the report before that moment and so would be reading it at the point when they will need to explain the findings to their patient. Two providers commented on the burden they imagine providers feel when they get a result on a condition that they know nothing about with reference to "*looking like an idiot*," as stated by this provider:

> *"You just think about . . .it takes the burden off the clinician to have to go find all of this material themselves and they don't have to look like an idiot the first time they're talking to the patient."*

The report section that included tables with prognosis, clinical guidance and next steps were repeatedly highlighted as extremely valuable by the participating providers. Several providers were particularly appreciative of the format that concentrated everything in one place, the temporal presentation of diagnostic and prognostic findings relative to the genomic result, and the comprehensive detail; stating:

> *"I love it—no clicks, detailed, comprehensive enough that I didn't feel I needed another source—everything a pediatrician would think about."*

> *"I think this is most important for me to know—what lies ahead and when to expect what, what I should do—glad it is broken down by time." . . .your confidence, your anticipation is right, because I don't know this disease."*

> *"This is great for someone who doesn't know much about rare conditions. I love idiot-proof stuff. Our knowledge of these things is miniscule, . . . that really lets you know in terms of symptoms,. . .tests that you can do, things you need to be looking out for, what you can do. Wow, this is great. I think this is awesome."*

However, two of these providers indicated concern that there was no mention of life expectancy in this information. This was especially important for one provider, with Internal Medicine training, regarding the incidental finding:

> *"I would be concerned about a primary finding that suggested early death—why are secondary findings important? Do we know that they will apply for this patient?"*

Overall providers overwhelmingly expressed the workflow improvement that could be realized through the use of the information in these sections of the genome report:

"...all about workflow: a way to hyperlink from the problem list, to [genomic results,] to treatment options ... going to really allow that flow to happen much easier than rely on someone to go and look for the genetic results and then Google ...and then hopefully find a website that has enough information that breaks it down the way they can digest it. [This way], by having everything hyperlinked in Epic where you can click on ...the problem list or genetic results and it is all there ...allow [providers]to at least think about the optimal care at the time."

The biggest challenge from the view of one provider had to do with *"what do I need to do for the patient in front of me and what do I have to give to the caregiver when they leave my office?"* Therefore, he proposed a plan for the next steps section that included converting it to a care and treatment plan checklist. He suggested developing smart sets with the choice available for referral—with a check box for the provider to indicate when the task had been completed.

The Genomic Report Can Serve as a Resource to Facilitate Communication of Genome Sequencing Results with Their Patients and Families

The provider participants consistently expressed that the design and information in this genome report could be used to improve or facilitate interaction and communication with their patient. The section that providers found most striking was the **Prognosis** section designed using the SimulConsult® diagnostic decision support system that included tabular presentation of typical condition-specific findings over time. This section was viewed as not just helpful for the clinician's understanding of the condition but as a useful communication tool for conveying this information to the family:

"if somebody can present ...that this [finding] is what is likely to happen to that particular patient, then I would focus on the [items that are seen] *most and say* [for example] ... *that there will be developmental delays and in most people you notice that beginning at 6 months. ..."*

"I would use it as communication...here's where we are today ... the [age] *intervals used are the same as I am used to looking at ...*[this is a] *good summary/synthesis of information..."*

"[this table is an]... *excellent, helpful way to get at the temporal aspect and ...points out that not every child has to have every finding. This helps in family communication about what* [the family] *need to worry about or not..."*

Providers also noted that the **Next Steps** section would help them to frame information about the findings and would support conversation with the parents about disease progression and care:

"I would pick it up when I'm having a conversation. I'm on the same page with you [patient] *and this is what we'll look for as a primary care* [provider]."

"It helps ...to sit at the table and talk about prognosis and allows you to ask the right questions about what may be happening. ...whatever it may be ...have a frank conversation with the parent to let them know [their child] *is progressing; ...placed out on a nice easy table and then I can go click look at and say all right where are we at today."*

The **Resources** section with hyperlinks to patient support groups and family organizations was also identified as facilitating communication by helping the provider give the patient specific assistance.

"...helps clinician identify rapidly ... when I bring the patient in to know what to tell them and offer them resources—that's above and beyond what other places are doing"

The information presented in the report regarding **Secondary** (Additional) **Findings** was also noted as useful for facilitating communication with the family and implications for relatives. These findings often affect the whole family and not just the child.

"I know the BRCA1 and 2 [risks] for females but knowing what to look for in males is going to be very important because it may make you do a much better discussion for prostate cancer in the future. ... children don't even worry about it, but as he gets older, this is what you need to do from a guidelines standpoint ...that kind of layout for the pediatric end, too, increases your yield for improved care."

However, while they expressed they would communicate the incidental result to the patient and family, all providers indicated that they would refer for genetic counseling with regard to the specific incidental finding of a *BRCA1* pathogenic variant. A few providers indicated that they might not refer their patients to a genetics professional for every incidental finding, as there may be some findings that they feel comfortable discussing with their patients. Several providers requested that the report provide referral information for genetics specialists.

"The BRCA associated cancers, ... this adds a lot of additional work for us. So is there a way of saying 'recommend the following: genetic counseling."

"I want to know who to refer to, but I also appreciate having some explanation so I can talk with the patient when the report is reviewed."

Regarding incidental findings, the providers also expressed concern for needing to communicate the importance of the result for other members of the family, not just the child who was tested. One provider had concerns about how best to communicate the

increased risk of cancer for the parent who carries the *BRCA1* variant. The provider suggested that the importance of this information needed "*to be worded strongly enough*" to influence their patient's willingness to be tested and receive care. This provider also asked for the information on which parent carried the variant if possible as a means of more directly facilitating this conversation.

Provider Opinion of the Patient-Facing Report

At the end of the semi-structured interviews, providers were given the opportunity to review the family genome result report. All providers were enthusiastic about having access to the report that could go to parents and indicated that they could see parents using the report in multiple situations:

> "*I mean parents would like pin this to the kid's chest when they roll in [to see the doctor]*"

Most providers wanted access to the family report, some stating that the report included information that was not in their report.

> "*I might not know to read the patient report. I might not know that it's different from, like very different from an information standpoint, so unless it is clearly said that there's additional information. Maybe a line at the end of the report saying this is a clinician version, there is a patient version . . .*"

> "*Does the clinician get this too? This would be really helpful to know what they're getting because when they come in, I want to be ready for these questions.*"

Another provider remarked on the information included in the family genome report addressing risk related to the *BRCA1* pathogenic variant and its relevance to the provider genome report,

> "*I think it should be there [in the clinician report]. I think the physicians should it have because I don't know off the top of my head. . . definitely pictures and tables are a better way for presenting it,.. I would use this page.*"

Providers were asked if the family genome report would be sufficient for both their purposes and the family's since the information was similar. However, providers unequivocally expressed their desire for their own report, but with access to the patient report.

DISCUSSION

In this study we collected provider responses regarding review of a genomic report generated to convey results of whole genome sequencing. The genome report contained fictional variant results pertaining to primary and incidental findings for a 10-year-old male with undiagnosed muscle weakness. None of the providers had experience with the primary condition or the associated gene finding. In addition, none of the providers had experience with communicating the incidental finding which involved a *BRCA1* pathogenic variant. In many respects this leveled the playing field among the various providers irrespective of their main practice setting. We propose that this is representative of the anticipated situation that many providers will experience as genomic testing gains momentum in the clinic. Providers will receive genomic results with new unrecognized rare gene variants and in addition may encounter incidental variant findings in known genes, but with which they may have no experience.

When participants were asked to review and comment on the genome report, each compared the report to their previous experience with laboratory reports. Providers preferred the genome report created for this study. In particular, many commented that the format and content of this report facilitated their understanding of the result and informed subsequent consideration of medical management. Their responses reflect attitudes described in previous studies that show that healthcare providers without experience in genetics consider genetic test results complex and laboratory reports hard to interpret [McLaughlin et al., 2014; Dorschner et al., 2014]. Providers identified that the section with a description labelled 'clinical rationale' facilitated greater understanding of the genome finding. In our report the clinical rationale section comprised an explanation of the gene, gene function and its potential clinical role in clear, concise terms.

Providers also appreciated that links to in-depth information were merely one click away. They indicated that they imagined they would feel more confident knowing that specific high quality resources were readily available to better understand and explain a genomic finding. All of the providers referenced that genome sequencing is different than ordering a single gene test where testing involves a known gene associated with an expected syndrome. However, with genome sequencing, they may know little or nothing about a gene or a variant, while still needing to explain it to their patient. Providers expressed frustration when they do not know what to say to families. The addition of supporting information about the nature of the gene and its potential clinical effect was also seen as enabling providers to better explain the results during the clinic visit. As we look to more fully integrate genomics into healthcare, there is need to develop innovative approaches to advance understanding of genome sequencing results within the point-of-care context. In this report we sought to deliver access to a broad range of genomic resources specific to the result. Some indicated that there might not be time during the patient visit to link out of the medical record, however, having the links available relieved their anxiety about being confronted with results that they knew nothing about. Providers highly endorsed the format and content with hyperlinks to disease specific information which could increase their understanding of genome sequencing results associated with unfamiliar and rare conditions.

The most valued feature of the report involved its potential to facilitate communication with patients and their family members about a rare diagnosis. Perhaps not surprisingly, providers were enthusiastic about the same sections as the patient participants. Provider comments mirrored patient comments on the value of a table that gave temporal presentation of prognosis. The Prognosis Table©, as developed through SimulConsult® and modified as a result of feedback from the patient participant interviews, was seen as a teaching tool for the providers themselves as well as with

patients or parents. The temporal presentation of the Prognosis Table©, was seen as particularly valuable. All remarked that the data was presented in a way that was very familiar and matched other review formats such as the Denver Developmental Scale which made it intuitive to Pediatricians. They described the report as offering a way to focus the conversation and structure expectations over time relative to the condition. They recognized the benefit of seeing language that explains the findings in simple terms. This was most apparent when the providers had the chance to review the patient version of the genome report. The providers requested access to both versions explaining that they did not want patients to have more information than they had. Providers expressed concern about patients asking questions to which they would not know the answers. As the interviews continued, it became apparent that this might be characterized using an economics descriptor known as information asymmetry, where one participant has more or better information than the other participant [Aboody and Lev, 2000]. They described the language, explanations and illustrations in the family report as appropriate for patients, but also helpful for themselves. The providers were satisfied when they learned that both reports can be viewed from within the patient electronic health record.

A difference between the Pediatric and Internal Medicine providers became apparent in review of the Prognosis Table©. While all saw tremendous value to the table, the Internal Medicine clinicians were dismayed that the information did not include reference to life expectancy, reproductive risks or other transitional issues that they often face with patients. They questioned the value of conveying incidental findings if the primary diagnosis is known to be associated with shortened life expectancy. In further discussion they proposed that there might be value around provision of incidental findings information to at-risk family members. The issue of whether to present as well as how to present information regarding life expectancy would benefit from further study with patients and family members. It will be important to learn what patients, parents and family members consider important about life-expectancy information and whether or how it should be conveyed.

One of the interesting exercises that many providers entertained during their interview involved imagining how they might use the next steps section to populate their clinic note or to generate a checklist for referrals and/or orders to ensure appropriate follow-up care for their patients. The idea that the report would be in the patient's EHR led them to think of a variety of potential important improvements in their clinic workflow beyond the fact that the report included the genome result and useful clinical information. They commented on the possibility that such a genome report could support their clinical care decisions by linking them with best care practices if available and providing a view into the future that would help with anticipatory guidance. Development of this functionality could increase the utility of the report for the providers.

Another major theme conveyed repeatedly across the various provider interviews involved use of the genome report as a tool in the communication of genome sequencing results. Just as providers were concerned about what the result was and what it meant, they were very concerned about getting the communication correct. This appeared most obvious as they discussed communication of an incidental finding. Many remarked that the issue of incidental findings was out of their practice experience and comfort. All physicians indicated that they planned to refer to genetics professionals for counseling about the incidental findings. However, they also appreciated that the report included brief information about management and risks. Most admitted that they were unlikely to review a result before the patient visit and so ran the risk of appearing uninformed about the primary finding and the incidental finding when they met with the patient to return genome sequencing results. The physicians indicated that they would be reluctant to convey such a result to patients if they knew nothing substantial about its implications. The providers endorsed that information on findings and management recommendations related to incidental findings were included in both the provider and patient versions of the report.

When no causal variants were identified to explain the patient's clinical findings, providers had concerns about how go back to the sequence over time. They struggled with the fact that the sequence was there and we might not know what to look for at this time. In our example report, we included a section that listed variants considered potential suitable targets for research. The inclusion of a list of variants generated a certain amount of internal conflict about whether or not to include the list. None wanted to miss something that might be found to be of importance in the future, but they also did not want the responsibility to go back to look and assess value of any given variant on the list. All eventually came to recommend that creating a formal re-analysis service, using the original sequence but with updated annotations and updated clinical discoveries may be important. Geisinger will be well suited to evaluate process options for clinicians because of the re-analysis capability now available through SimulConsult®.

Limitations

This study did not capture response to a truly web-enabled genome results report. Providers reviewed a paper version of the result report and were asked to imagine the ability to click-out to resources. The practical reality of using the report with the workflow of the visit context remains untested. Only six providers were included in this study; however, saturation was reached. This may be explained because the providers had at least some experience with genetic reports, thus, testing the report with providers who are unfamiliar would be an important next step. This work will continue with additional providers, and iterations of this draft will provide us with additional insight.

The next step in this study is to deploy the web-enabled enhanced genome reports for both providers and patients in a prospect comparative effectiveness trial, registered at clinicaltrials.gov, evaluating patient satisfaction and communication. Half of the patients (and their caregivers) will be randomized to receive the enhanced report to communicate genome sequencing results via the patient portal and half will receive usual care which consists of communication of sequencing results via a visit summary letter. At six months those in usual care will receive the enhanced genome report allowing additional analysis using a cross-over design. Providers identified by the patients/caregivers will receive access

to the provider report simultaneously allowing study of changes in communication between the two groups attributable to the report.

CONCLUSION

Initial testing demonstrates the desirability of a report designed to convey results of whole genome sequencing including primary and incidental findings. Critical elements of the report included clear language to explain the result, as well as consolidated yet comprehensive prognostic information with clear guidance over time for the clinical care of the patient. Most importantly, it appears a report with this design has the potential not only to return results but also serves as a communication tool to help providers and patients discuss and coordinate care over time.

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