Communication challenges for nongeneticist physicians relaying clinical genomic results

Aim: Identify the behavioral challenges to the use of genome sequencing (GS) in a clinical setting. Materials & methods: We observed how general internists and nongenetic specialists delivered GS results to patients enrolled in the MedSeq Project. Using transcripts of such disclosure interactions, we made qualitative observations of communication behaviors that could limit the usefulness of GS results until reaching the point of thematic saturation. Results: Findings included confusion regarding genomic terminology, difficulty with the volume or complexity of information and difficulties communicating complex risk information to patients. We observed a broad dismissal of clinical value of GS by some physicians and sometimes ineffective communication regarding health behavior change. Conclusion: Overcoming these behavioral challenges is necessary to make full use of clinical GS results.

First draft submitted: 10 February 2017; Accepted for publication: 16 May 2017; Published online: 15 September 2017

Keywords: behavior change • clinical genomics • ELSI • genetic literacy • physician–patient communication • qualitative research • whole genome sequencing

Background

Many genomics experts believe that whole exome and whole genome sequencing, heretofore referred to as genome sequencing (GS), hold great promise for improving healthcare and patient outcomes [1,2]. GS has been shown to be effective in diagnosing rare genetic conditions in a substantial proportion of cases, ending for many patients what has been a long diagnostic odyssey [3,4]. However, the promise of personalized medicine extends beyond the diagnosis of monogenic genetic disorders to a world in which everyone can potentially benefit from GS. For example, some suggest that population-wide GS could optimize preventive healthcare strategies and drug therapies [2,5], based in part on evidence that 1% of the US population may have a highly penetrant mutation for which a preventive strategy is available [6]. Others suggest that GS could promote health behavior change because genomic information is more personalized than general risk information [7]. Experts also point to the potential value of pharmacogenomics to determine which medications at which doses should be prescribed for specific patients, to achieve therapeutic response and minimize side effects [8]. Finally, some experts predict that GS will aid couples in family planning decisions through the identification of autosomal recessive conditions [2,9]. For GS to realize these promises, genomic science must continue to advance. It is also acknowledged that the costs of GS, including the costs of interpretation, need to further decline for such results to meaningfully contribute to routine clinical care [10].

However, less often recognized are the behavioral barriers to the use of GS in clinic settings [11]. Many have acknowledged that, if GS becomes part of routine clinical care, there are not enough genetic specialists to help communicate GS results [12,13].
Yet, primary care physicians and nongenetic specialists report that they are not comfortable communicating even single gene test results to their patients [14,15]. If GS is to improve clinical care for a wide variety of patients, then primary care physicians and nongenetic specialists will need to first understand and then be able to explain GS findings to their patients; moreover, patients need to understand the GS information to incorporate it meaningfully into their health management.

As investigators in the MedSeq Project, a randomized trial comparing GS to standard of care (family history), we set out, among other things, to determine whether primary care physicians (PCPs) and cardiologists (after a 6-h education session on genomics) [16] could appropriately disclose GS results without a genetic specialist present. Some of our results have been quite promising. In previous analyses, for example, we showed that the majority of primary care physicians were able to competently understand and communicate monogenic disease risk variants to their patients, as judged by a panel of clinical geneticists, without making significant errors or contributing to safety or malpractice concerns [17,18].

But in observing how physicians discuss GS results to patients, we also came across some more troubling communication behaviors that potentially limit the usefulness of GS results to patients. In this article, we present additional qualitative observations from the study, specifically evaluating ways that some PCPs and cardiologists delivered whole genome sequencing test results to patients that potentially undermine the value of that information to patients. These observations were made by reading transcripts of interactions between physicians and patients, in which they disclosed GS results. Although we observed many examples of effective communication, here we focus only on communication behaviors that could undermine the value of GS findings, with the goal of helping clinicians develop ways to improve upon these behaviors.

Materials & methods
Overview
This analysis is part of the MedSeq Project, whose protocol is described in detail elsewhere [16]. In brief, the MedSeq Project is a randomized controlled trial comparing GS to current standard of care (family history) in two clinical contexts: disease-specific genomic medicine in a cardiomyopathy clinic and general genomic medicine in primary care. In this study, the physicians were also research participants and both physician-participants and patient-participants were consented as part of an IRB-approved protocol. We enrolled eleven primary care physicians and nine cardiologists from a large urban network of academic hospitals and outpatient practices. A total of 100 adults were enrolled in the primary care cohort and 102 adults in the cardiology cohort. Physician-participants were provided a 6-h educational curriculum and offered a hotline to genetics professionals for guidance in interpreting their patients’ genomic information. A 5–6 page genome report was provided to physicians for each of their patient’s genomic results [19]. The report featured a single-page summary of results of potential medical relevance. Additional pages were devoted to structured variant, gene, and disease information along with supporting evidence for reported variants and brief descriptions of associated diseases and their clinical implications [19]. A copy of a sample report is available as an online supplement. In this analysis, we focus on the interactions between both groups of physicians and their patients who participated in the study in which the physicians disclose GS results.

Framework for communicating GS results to patients
To guide our analysis of the disclosure interactions, we utilized a shared decision-making model to assess the communication of GS results by physicians to patients [20,21]. In our ideal model, the patient is first counseled on potential risks and benefits of GS prior to genomic testing. Once the patient’s genome is sequenced, the report is returned to the physician. Appropriate communication depends upon the physician understanding the information. Following that, if clinically useful information is available from sequencing, either the physician relays the information to the patient and recommends that the patient make a behavior change (e.g., recommending more exercise) or the physician directly alters the patient’s treatment plan (e.g., changing the dose of a medication). Alternatively, the finding may be purely of informational value for the patient (nonactionable), or there may be no clinically useful information to return to the patient. In this analysis, we identify specific physician behaviors that have the potential to reduce the effectiveness of this communication. For example, deviations from the ideal may occur if physicians do not understand the genomic information, are confused by the terminology, or are overwhelmed by the volume of information.

Qualitative content analysis
We read transcripts of interactions between physicians and patients in which physicians disclosed GS results to patients and noted instances in which the communication deviated from the model. After reading a number of transcripts, the team re-read the portions of
transcripts identified as deviations to form discrete categories of communication challenges. We continued collecting and deliberating upon case examples until we reached a point of thematic saturation [22], whereby subsequent deviations were a result of behaviors we had already categorized.

Our multidisciplinary qualitative team was made up of two researchers experienced in analyzing physician/patient interactions, an undergraduate student studying genomic sciences and policy, and an experienced physician with expertise in shared decision-making.

Focus on behavioral factors
We specifically identified physician behaviors that potentially stand as barriers to maximizing the clinical value of GS testing. We excluded nonbehavioral factors from this analysis, such as technical and clinical limitations of GS. For example, some physicians explained to patients that the clinical significance of certain GS variants was unknown. Yet at other times, physicians pointed out that the risk information provided by the GS report was too vague to act upon, such as pharmacogenomic results indicating intermediate risk of developing side effects to a particular drug, with no information on what constitutes ‘intermediate risk’.

We excluded conversations about these issues from our analyses because they represent potentially surmountable scientific limitations of GS, and our analysis was focused on behaviors that stand in the way of incorporating more useful GS results into clinical care.

We also excluded conversations that revealed clinical limitations of GS data. For example, physicians sometimes pointed out that pharmacogenomic findings were not relevant to a given patient, because the patient had no foreseeable need for the medication in question. At other times, physicians pointed out that GS results were not relevant as predictive information, because the patient already had phenotypic data or the trait in question, such as when a physician pointed out that a patient’s “HDL is much better than” the GS test predicted and “so this doesn’t shed any light on your situation”.

In addition to reporting specific behaviors of physicians that are potential barriers to adoption of GS, we identified some of the difficulties that physicians have motivating patients to make behavior changes. Ideally, physicians would utilize scientifically validated methods of encouraging behavior change, such as motivational interviewing and assessing patient’s readiness for change through a stage of change model [23,24]. However, we identified situations in which physicians use a more directive, and likely less effective [24,25], way to promote behavior change. These communication behaviors are not specific to genomics; physicians often struggle to motivate patients to adopt healthy behaviors [26,27]. However, given what some think is the potential for genetic information to be an additional motivator for patients to change their behaviors [28–30], it is important that physicians utilize effective tools to motivate behavior change in the context of GS.

Results
We identified communication behaviors that affect how physicians adopt GS into clinical care (Table 1). Several of these behaviors are applicable in realms beyond genomics and are areas of improvement for all physicians more broadly.

Physicians may be confused regarding genomic terminology
In order for physicians to effectively communicate GS findings to patients, they need to be able to understand the findings and reports. However, several factors can prevent their full understanding of genomic information. First, they can lack understanding of genomic terminology. For example, some physicians became lost in the letters and numbers of the genomic code while interpreting the results for patients. In one interaction, the physician told the patient that he was “intrigued by this variant classification”. He continued, explaining that “they know the exact position of the gene” and says “that’s what the mumbo jumbo-C5775 – but heterogenic genus [sic], and then they said likely pathogenic. That’s the question I have. What does that mean?” In this interaction, it appears that the physician does not understand the genetic information well enough to explain it to the patient, and they both leave the conversation confused. In another encounter, the physician told the patient with obvious irony, “We’ll both read about methylmalonic aciduria and homocystinuria. Right?” Then, they both laughed. In both instances, by conveying lack of understanding of the genomic information and then presenting jargon to the patient, the physicians have not succeeded in translating the information into a form that helps patients use the information.

Physicians may be overwhelmed by volume or complexity of information
In addition to difficulty with terminology, physicians were sometimes overwhelmed by the volume of information provided to them in the report. As one physician expressed to a member of the study team before disclosing information to study participants: “there can be such a thing as too much information.”

In addition to the volume of information, physicians may struggle to comprehend the information because it is complex. As one physician explained to a
patient: One thing I struggle with is the complexity of this information and how possibly people are going to interpret – the layperson is going to interpret it when they spend their thousand bucks. Above average could – someone could lose their sleep over above average.

Another physician complained of not having enough time to process GS results in advance of meeting with the patient: “I don’t know if I had an opportunity to actually sit down and go over this with somebody smarter than me. Actually, I think the answer is I did not have an opportunity. I get this just a few days before you come here.”

Others indicated this complexity indirectly, by jumping between thoughts, conditions and expressing a lack of confidence in their ability to understand specific topics and conditions.

If physicians are struggling with genomic information for any of these reasons (jargon, volume and complexity), they may experience difficulty effectively explaining what the information means to their patients. It is important to recognize that these physicians had such challenges even after participating in a 6-h orientation to the reports and the material given by members of the MedSeq Project team on understanding and interpreting genomic findings [16]. Notably, this educational session did not include information on how to communicate genomic results to patients; it only provided education about basic concepts in genetics. Physicians also had an opportunity to consult with a genetic specialist on the study team at any time about their patients’ results. These challenges could be exacerbated for physicians who do not receive such training or have such a resource available to them.

Physicians may struggle communicating complex information, especially related to risk

The way that physicians present genomic information to patients can also hinder patient understanding. Even if the physician understands the GS results, if she is not able to relay the information in clear terms, the patient may not understand. In some disclosure sessions, physicians presented lengthy, technical explanations of information without verbally assessing for patient understanding. This problem is not unique to genomics, but could be amplified in the setting of new, complex and uncertain information. One physician explained:

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<th>Table 1. Communication challenges for nongeneticist physicians relaying clinical genomic results.</th>
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<td>2. Physicians confused regarding genomic terminology</td>
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<td>3. Physicians overwhelmed by volume of information</td>
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<td>4. Physicians broadly dismiss value of WGS</td>
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<td>5. Physicians inadequately motivate patients to make behavior changes</td>
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EKG: Electrocardiogram; HCM: Hypertrophic cardiomyopathy; WGS: Whole genome sequencing.
So this part here is all about statistical – the statistics and chances and ratios and you know. So because – so that’s what we’re going to go over. So let’s say we’re going to do hyper – so why don’t I start saying that these are more about the statistics and probabilities than – so it’s not so determinant as the other things that we talked about, and you’re going to see it very clearly.

The physician’s explanation here is, arguably, confusing, making it concerning that she does not verbally assess for patient comprehension. She further explained that because the patient had 74 copies of genes associated with diabetes, he had a 3.5-fold increased risk of developing diabetes compared with a 'regular person', with no mention of what this risk means for the patient in absolute terms or in the context of the patient’s life. From the patient’s lack of response following the physician’s comments, it is unclear whether he understood the information.

**Physicians might broadly dismiss value of GS**

Several physicians demonstrated a negative bias toward the value of GS with some of their patients. The broad dismissal of the value of genomic sequencing by physicians can affect the perceived utility of the test. One physician explained to a patient that ‘most useful’ information from the study was from the family histories, rather than the GS test. In another disclosure session, a physician emphasized that genomic sequencing was ‘not ready for primetime’. Existing physician biases, even among this group of presumably proactive, open-minded physicians, can lead to comments that influence the downstream perceptions and opinions of their patients. The dismissal of value of genomic testing by physicians can decrease the likelihood that patients will take the information that is presented seriously.

**Physicians may fail to adequately motivate patients to make behavior changes**

Given that some experts believe that GS results can be used to promote health behavior change because genomic information is more personalized than general risk information [7], we focused on instances in which physicians could have attempted to motivate patients to make behavior changes based on the genomic information. There are established techniques recommended to physicians for promoting patient health behavior change. These include motivational interviewing and utilizing the trans-theoretical model to assess the patients readiness for change [23]. In this situation, physicians could utilize the genomic information provided by the report to further encourage patients to modify their health behaviors [11].

However, many doctors used less effective strategies when trying to connect GS findings to the need for behavior change. For example, one physician told a patient that it is “very important for you to eat properly and exercise regularly. Those are the two things that you can do that would be most effective in preventing your risk of developing diabetes.” She did not go on to further explore any potential barriers to doing so for the patient. Another physician told a patient, “You can’t change your genetic makeup, but you can change your diet and your exercise.” When the patient simply responded, “Okay”, the doctor replied, “So that’s something you can work on” while doing nothing else to explore strategies for how the patient could achieve this goal. It is important to note that we are looking at these encounters as brief snapshots of the physician-patient relationship, and the physicians may have used other health behavior change techniques in other encounters with the same patients. However, if we consider genomic risk information to be one more tool to promote health behaviors as physicians, then using best practices for behavior change counseling would capitalize on this otherwise missed opportunity.

**Discussion**

In a study of interactions between physicians and patients, we discovered a number of behaviors that potentially limit the successful adoption of GS in clinical care. Importantly, we do not suggest that these behaviors are the norm when physicians disclose GS results; here we focused explicitly on less effective communication. Findings specific to physicians included confusion regarding genomic terminology, difficulty with the volume or complexity of information, and difficulties of communicating complex risk information to patients. In addition, other areas we explored included a broad dismissal of value of GS by some physicians and sometimes ineffective communication between physicians and patients regarding the need for health behavior changes.

It was not surprising that some physicians struggled with interpreting genomic findings for patients, given their relative lack of training in this area in comparison to genetic counselors or genetic specialists [14,31]. Though physicians explained the information to patients in a way that largely avoided errors that would compromise patient safety as previously noted [17,18], we highlighted potential areas of improvement. Additionally, some physicians may be more broadly dismissing the value of GS because at this point the clinical utility is not clear to them. This sentiment may change as the technology progresses. There is also disagreement within the scientific and medical community regarding which genomic information ought to be returned...
to patients [32,33], and it is important to acknowledge that this can lead to variation in how physicians choose to communicate genomic information. It is clear that regardless of which information physicians choose to disclose, they ought to communicate that portion clearly to patients. In order to fully realize the promise of genomic sequencing, primary care physicians and nongenetic specialists need to be more thoroughly trained on how to understand genomic information and how to use genomic results to motivate behavior change, or genetic specialists will need to be trained in adequate numbers to perform these tasks. To overcome some of the challenges that we have raised, we can continue to improve the design of clinical GS reports. The report designed by the MedSeq Project team sought to mitigate the challenges of overwhelming information for physicians by presenting information in a clear, concise manner [16,34,35]. Genomic reports that highlight salient clinical information can reduce the burden on physicians to understand every piece of information. Such reports could be integrated with the patient’s electronic health record to display the most relevant information. Well designed, clear, and succinct reports can also reduce the burden of dealing with high volumes and high complexity information for physicians.

We have previously discussed the ‘last mile problem’ in genomics, which would be the failure to integrate this new technology into society in a way that improves human behavior and decision-making [11]. Physicians will need tools from behavioral science to convince patients to make lifestyle changes [11]. Furthermore, others have explained how patients may anticipate a greater impact of genomic results than they actually experience, and we can counter this ‘impact bias’ by combining the reporting of genomic information with other techniques that are known to promote behavioral change [11,36,37]. Such tools include motivational interviewing, targeting interventions appropriate to the patient’s readiness for change, and employing immediate incentives that leverage present bias [23,24].

One way to overcome the ‘last mile problem’ is to better educate physicians, starting from medical school onwards, on how to communicate clearly with patients regarding genomic information, risks, and the potential for behavior change. Students are educated on behavior change models at some medical schools [38], and it is possible to include information on how to incorporate genetic information into such conversations into medical school curricula. Standardized patients may be a particularly useful pedagogical tool here.

The communication challenges we have identified in this analysis are not unique to genomics and exist across the spectrum of medical care [39]. For example, patients cannot make appropriate use of many medical tests — including laboratory tests and imaging studies — without understanding the meaning of the test and how to apply test results to their lives. When physicians struggle to communicate test result information to patients in a comprehensible manner, the same issues arise as we observed among some of the participants in the current study [39–41]. Nevertheless, communication challenges loom especially large for new and complex technologies, making these communication challenges of special concern for the successful use of GS.

Using basic technology available in many doctors’ offices as early as 2004 [42], it is possible to have online modules for patients to complete prior to a doctor’s visit so they can acquire basic genetic literacy and quell anxieties about what they may learn in their visit with the physician. Communication techniques relevant to any clinical encounter, such as showing an interest in patients’ ideas about their own health, allowing patients to talk without interruptions, and encouraging patients to ask questions, are equally important when discussing genomic health information [43].

Another area where physicians currently struggle is relaying risk information to patients. Often, responses to risk, such as fear and anxiety, are unduly influenced by the framing of risk rather than the likelihood of the risk. Therefore, telling individuals that they have an above average risk for a difficult disease could unnecessarily alarm them — even if that risk is minuscule in impact. The way that we frame and contextualize these risks will help patients better understand their clinical impact [44]. Some best practices for conveying risk information to patients include using plain language and presenting data using absolute risks. In addition, presenting data using frequencies and pictographs can be effective [44]. All of these methods can be utilized in preparing the genomic reports as discussed earlier. It is important for physicians to realize that comparative risk information is persuasive and not just informative [44]; thus, it is not advisable to tell patients their risk is 3 times higher than usual if that risk changes from 0.1% to 0.3%. Other areas for future investigation may include determining whether physicians are able to adequately counsel patients on the differences between risks that directly affect them and risks that affect their carrier status for a particular disorder.

Our study had several limitations. First, we did not attempt in this report to give a balanced sense of the communication between physicians and patients. We explicitly looked for examples of less effective communication. Readers should understand
that there were many instances in which physicians counseled appropriately, but they were not included in our analysis. We do not aim to suggest that the behaviors we document are the norm, even for this limited sample of physicians and patients. Instead, we sought to identify suboptimal behaviors for the purpose of highlighting, categorizing and overcoming those challenges. Second, because we observed the physician-patient relationship for one encounter, we do not have a full view of how the interactions we assessed stand in the context of the long-term relationships between physicians and patients. As a result, we do not know if these communication behaviors impacted patient outcomes or clinical utility. Third, our study is qualitative and we did not address how often each of these behaviors occurred. In part, we avoided quantification out of recognition that there is not an exact threshold between effective and ineffective communication. Rather than quantify these behaviors, we sought to characterize these phenomena and present with clear examples that could serve as lessons. Furthermore, the issues that we present are not unique to genomics. Communication behaviors impact all fields in medicine. The challenges physicians face in the context of genomics may be amplified because the information is often newer, less understood, complicated, and uncertain.

Finally, though we have focused on situations in which physician behaviors limit the usefulness of GS, there were many instances in which physicians relayed genomic information well to patients. For instance, some physicians explained the current state of GS and potential growth without being dismissive and some were able to adequately convey risk information using absolute rather than relative terms. Some physicians used prefacing statements to put patients at ease, such as one physician who explained to a patient that while the reports may have a lot of information, together they would break down the information into individual components to make it easier to understand. In another interaction with a patient, a physician discussed a referral to a genetic counselor who could better answer some of the patient’s questions. Overall, some physicians communicated genomic information in a deliberate, clear, and succinct fashion. As we have previously described, in this study we focused on communication challenges to help identify areas for physicians to improve. Future studies could analyze communication behaviors of nongeneticist physicians who relay genomic information effectively to provide insight into optimal communication patterns.

**Conclusion**

Ultimately, it is important to recognize that the physician behaviors presented here are surmountable challenges. Confusion regarding genomic terminology and challenges in relaying risk information can be addressed through physician education. Strategies for incorporating genomic information in health behavior change conversations can also be taught. Continued improvements in the design of clinical GS reports can allay difficulties with the volume and complexity of the information. Moreover, physician attitudes toward the utility of genomics may change with familiarity and demonstrated utility. With proper training and support, physicians can come closer to the goal of using GS to bring the right clinical care to the right patients at the right time.

**Summary points**

- While whole exome and whole genome sequencing (GS) have the potential to improve patient care, the behavioral challenges to the use of GS in a clinical setting are not often recognized.

**Methods**

- We observed how general internists and nongenetic specialists delivered GS results to patients in the MedSeq Project.
- Using transcripts of interactions in which physicians disclosed GS results to patients, we made qualitative observations until reaching the point of thematic saturation.
- Though we observed many examples of extremely effective communication, here we focus only on communication behaviors that could potentially limit the usefulness of GS results.

**Results**

- Findings among physicians included confusion regarding genomic terminology, difficulty with the volume or complexity of information, and difficulties communicating complex risk information to patients.
- In addition, we observed a broad dismissal of clinical value of GS by some physicians and sometimes ineffective communication between physicians and patients regarding health behavior changes.

**Discussion**

- To fully realize the promise of genomic sequencing, primary care physicians and nongenetic specialists will require training on how to understand and communicate genomic information and how to use genomic results to motivate behavior change.
Acknowledgements
The authors thank the staff and participants of the MedSeq Project for their important contributions. The authors thank 5AM Solutions, Inc. (Rockville, MD, USA), for their help in customizing the work flow of the ‘My Family Health Portrait’ web tool for this study.

Financial & competing interests disclosure
The MedSeq Project is carried out as a collaborative study supported by the National Human Genome Research Institute U01-HG006500. Dr. Robert C Green receives compensation for speaking or consultation from AlA, GenePeeks, Helix, Illumina, Ohana, Prudential and Veritas; and is co-founder, advisor and equity holder in Genome Medical, Inc. The authors have no other relevant affiliations or financial involvement with any organization or entity with a financial interest in or financial conflict with the subject matter or materials discussed in the manuscript apart from those disclosed.

No writing assistance was utilized in the production of this manuscript.

Ethical conduct of research
The authors state that they have obtained appropriate institutional review board approval or have followed the principles outlined in the Declaration of Helsinki for all human or animal experimental investigations. In addition, for investigations involving human subjects, informed consent has been obtained from the participants involved.

Important discussion of the cost–effectiveness of genomic medicine.


To further clarify the importance of behavioral research in advancing genomics.


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For further information regarding the design and layout of the MedSeq Project.


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Papers of special note have been highlighted as: • of interest;
•• of considerable interest
•• Description of shared community of research sites that is advancing evidence-based genomic medicine.
To learn physicians’ specific concerns in reporting genomic results

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