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Physician Communication of Genomic Results in a Diagnostic Odyssey Case Series

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Abbreviations: IDIOM: Idiopathic Diseases of huMan study

MCCS: Medical Communication Competence Scale
U.S.: United States
WGS: Whole genome sequencing

Table of Contents Summary (max 25 words; a brief insight into what the article is about; it should entice the reader to go beyond the table of contents page and read the full article)

This multi-perspective study aims to gain further insight into physician communication of patient genome sequencing information.

What's Known on This Subject (max 40 words, in paragraph style)

Research has largely taken a single-perspective (e.g., physician perspective or patient perspective) approach towards understanding genomic results-related communication. Moreover, previous findings suggest that as a group, physicians face challenges in effectively communicating patient genome sequencing information.

What This Study Adds (max 40 words, in paragraph style)

This study describes a multi-perspective research approach to study physician communication of patient genome sequencing information in diagnostic odyssey cases. Findings suggest that physician communication of patient genome sequencing information is suboptimal compared to communication of general patient medical information.

CONTRIBUTORS' STATEMENT PAGE

Caryn Kseniya Rubanovich, M.S.—drafted the initial manuscript, conducted statistical analysis and interpretation of results, contributed to data management and quality control.

Cynthia Cheung, M.A., M.P.H.—drafted parts of the initial manuscript, critically reviewed and revised manuscript for intellectual content, conducted statistical analysis and interpretation of results, contributed to data management and quality control.

Ali Torkamani, Ph.D.—contributed to interpretation of results, critically reviewed and revised manuscript for intellectual content.

Cinnamon S. Bloss, Ph.D.—conceptualized and designed the study, selected assessment instruments, coordinated and supervised data collection, conducted statistical analysis, contributed to interpretation of results, critically reviewed and revised manuscript for intellectual content.

All authors approved the final manuscript as submitted and agree to be accountable for all aspects of the work.

ABSTRACT

Background and Objectives: The availability of whole genome sequencing (WGS) is increasing in clinical care, and WGS is a promising tool in diagnostic odyssey
cases. Physicians' ability to effectively communicate genomic information with their patients, however, is unclear. This multi-perspective study assessed physicians' communication of patient genome sequencing information in a diagnostic odyssey case series.

Methods: We evaluated physician communication of genome sequencing results in the context of an ongoing study of the utility of WGS for diagnosis of rare and idiopathic diseases. A modified version of the Medical Communication Competence Scale (MCCS) was used to compare patients' ratings of their physician's communication of general medical information to communication of genome sequencing information. Physician self-ratings were also compared to patient ratings.

Results: A total of 47 patients, parents, and physicians across 11 diagnostic odyssey cases participated. In 6 of 11 cases (54%), the patient respondent rated the physician's communication of genome sequencing information as worse than general medical information. In 9 of 11 cases (82%), physician self-ratings of communication of genome sequencing information were worse than the patient respondent's rating. Identification of a diagnosis via WGS was positively associated with physician self-ratings ($p = .021$), but not associated with patient respondent ratings ($p = .959$).

Conclusions: These findings suggest that even in diagnostic odyssey cases, where genome sequencing may be clinically beneficial, physicians may not be well-equipped to communicate genomic information to patients. Future studies may benefit from multi-perspective approaches to assess and understand physician-patient communication of genome sequencing information.

Word Count: 250

INTRODUCTION

The utilization of genetic and genomic technologies is expanding in the practice of medicine [1]. The relative dearth of genetic counselors and medical geneticists in the United States (U.S.) [2-4], suggests that physicians may be increasingly faced with interpreting and communicating genomic test results to their patients [5-8]. In spite of this, many physicians do not feel well-equipped to carry out the various aspects of genetic and genomic testing [9-13], such as ordering genetic tests [14], utilizing the genetic test results [15], and providing explanations of genetic test results to patients [7, 16-18]. Lack of education and training in genetics and genomics [7, 17, 19-29] has left physicians ill-prepared to interact with and communicate genetic results [9, 10, 17, 23, 25, 30-32] as medical curricula have not kept pace with genomic advances and the integration of these advances in clinical settings.

The potential for genetic and genomic technologies to improve health outcomes is substantial, but it is also dependent upon the appropriate translation into medical practice [8, 26, 33, 34]. This is especially relevant in the field of pediatrics, as genetic disorders are common in this population and contribute to a sizeable proportion of admitted cases in inpatient pediatrics units [35, 36]. Given the highly specialized and complex nature of genetic and genomic data, particularly in these types of cases, communication with patients about these data is often challenging and results may be difficult for patients and families to understand [37, 38]. Additionally, the ways in which physicians' share this information may impact a patient's or parent's ability

to grasp the information and may reduce the overall quality of medical communication [39, 40]. For example, concerns have been raised as to how, if at all, physicians would use genomic information [41], and whether physicians can effectively communicate genomic information during a standard 15-minute clinical interaction [42].

There have been several attempts to understand barriers to productive physician-patient interactions about genomic information. Whether in commentaries [4, 8, 11, 12, 24, 26, 40, 43], systematic reviews [10, 17], surveys [7, 9, 44], qualitative work [18, 25, 45-50], or essays [23, 51], previous work has largely focused on the perspectives of physicians [4, 7-10, 12, 17, 18, 23-25], patients [44, 45] or parents [46, 47, 49, 50], but rarely on the perspectives of two or more of these groups simultaneously [48, 51-53]. In practice, however, the sharing of genetic test results occurs within the context of a clinical interaction between physicians and patients, or between physicians, patients, and parents. Thus, we have aimed to help address this gap by adopting a multi-perspective approach to better understand these real-world occurrences.

The current study aims to understand physician-patient and physician-parent communication through a series of 11 case studies of diagnostic odyssey patients who underwent whole genome sequencing (WGS) and the physicians who agreed to return genomic results. Specifically, we assessed both physician and patient/parent perspectives of the same genomic results-related communication and compared these different perspectives. Case series studies can help gather descriptive information and generate

hypotheses for future research on novel clinical phenomena [54]. Thus, we have employed this methodological approach to gain insight into the communication between physicians, patients, and/or parents that is related to return of patient genome sequencing information.

METHODS

Data from this study were collected as part of the Idiopathic Diseases of huMan (IDIOM) study, an ongoing initiative led by investigators at the Scripps Translational Science Institute. This study was approved by the Scripps Institutional Review Board (IRB-11–5723). IDIOM aims to uncover potential genetic causes for rare and/or undiagnosed diseases using WGS [55], and prior work in the context of IDIOM has also explored patient perspectives on participation in the study using qualitative methods [45].

Participants

Adults and children with a wide range of severe, undiagnosed conditions (e.g., neurological, gastrointestinal, hematological) were recruited into IDIOM. Recruitment and screening procedures for the IDIOM study have been described elsewhere [55]. Inclusion criteria consisted of patients who had: (1) symptoms that were considered potentially actionable with appropriate treatment and intervention, (2) a condition likely genetic in its origin; and (3) a “physician champion” who agreed to work with the research team and took on the role of returning genomic results. As part of IDIOM, the patient and the patient’s biological mother and father underwent WGS, and the patient’s

physician was responsible for returning the genomic results. As such, this study presents a unique opportunity to evaluate physician communication of patient genome sequencing results.

Procedures

This study utilized a pre/post-sequencing study design in which each participant completed an assessment battery. Depending on what was most convenient for the patient, about half of the participants completed a paper version of the battery and half completed an electronic version using SurveyMonkey. Ten different measures were included in the battery (see Supplemental Table 1), four of which were used at both pre- and post-WGS time-points. Here, we present results from two of these measures, a modified version of the Medical Communication Competence Scale (MCCS) [56] and a modified version of the Decision Regret Scale (DRS) [57]. The length of time required to complete the questionnaires was not explicitly tracked, but estimated to be an average of 15 minutes during each pre/post-sequencing time-point. Overall, questionnaires were completed between July 2012 and January 2014.

Patients and Parents

The MCCS (see Appendix A) assessed each patient's or parent's perception of the physician's communication of general medical information (assessed pre-sequencing), as well as the physician's communication of the patient's genome sequencing information (assessed post-sequencing) [56]. The

DRS (see Appendix B) assessed regret with respect to the decision to undergo WGS (assessed post-sequencing) [57].

In cases where the patient was less than 20 years of age (5 of 11 cases in our sample), one or both of the patient's parents completed these measures on behalf of the patient. In these cases, we used data from the mother's responses and considered the mother as the "patient respondent." We made this choice given literature showing that women tend to be the primary health care decision makers for their families [58] and are often the primary caretakers of their children. In cases where the patient was over 20 years of age, we used data from the patient's responses. Table 1 indicates the respondent for each case we studied.

Physicians

The MCCS was also used to assess each physician's self-perception of communication of the patient's genome sequencing information (assessed post-sequencing) [56]. The Decision Regret Scale [57] was similarly used to assess regret surrounding the decision to complete WGS for the physician's patient.

Measures

Medical Communication Competence Scale (MCCS)

This self-report scale was modified and administered both pre- and post-sequencing. Our MCCS is a 19-item instrument that uses a 7-point Likert-type scale, and response options range from strongly disagree to strongly agree, where respondents indicate their agreement with a series of

declarative statements [56]. The MCCS has shown adequate internal consistency (Cronbach's α = .75-.90) [56]. Statements on the MCCS were written from the perspective of the respondent such that a patient received a version written from a patient's perspective (e.g., *My physician…*) while a parent received a version written from the perspective of a parent (e.g., *My child's physician*…). See Appendix A for a copy of the modified MCCS that was used. Sample statements include, *My physician seems knowledgeable about my medical problems* and *My child's physician does a good job of reviewing or repeating important information*. At the pre-sequencing time-point, the patient respondent was asked to rate their physician's communication of general medical information. Only patient respondents completed the MCCS at the pre-sequencing time-point (Supplemental Table 1).

At the post-sequencing time-point, patient respondents were asked to consider the study-related clinical interaction focused on the physician's communication of genome sequencing information. Sample statements on the post-sequencing MCCS include: *My child's physicians did a good job of reviewing or repeating important information about my child's whole genome sequencing results* (patient item) and *I was comfortable discussing my patient's whole genome sequencing results* (physician item). Patient respondents and physicians both completed the post-sequencing MCCS.

Decision Regret Scale (DRS)

This self-report scale was modified and administered post-sequencing. The DRS assesses regret after a medical decision using a 5-item, 5-point Likert-type scale. Response options range from strongly disagree to strongly agree, where respondents indicate their agreement with a series of declarative statements [57]. The scale has shown adequate internal consistency (Cronbach's α = .81-.92) [57]. Respondents were asked to reflect on the decision to undergo WGS (in the case of the patient respondent) or participate in their patient's WGS (in the case of the physician respondent). See Appendix B for a copy of the modified DRS that was used. Patient respondents and physicians both completed the DRS post-sequencing.

Data analysis

All measures were independently scored and verified by two members of the research team. Data were analyzed using SPSS Version 24.0 (SPSS Inc., Chicago, IL, USA). Demographic characteristics were reported as frequencies and percentages for categorical variables. MCCS and DRS total scores were reported as medians. Measure reliabilities were calculated using Cronbach's alpha. MCCS scores from pre-sequencing and post-sequencing were compared for patient respondents, and MCCS scores from post-sequencing were additionally compared between patient respondents and physicians. These comparisons were performed using non-parametric Wilcoxon signed-rank tests due to non-normal distribution of the data and the presence of outliers. Post-sequencing DRS scores were also compared using a similar approach between patient respondents and physicians. In

addition, we used a Spearman rank correlation to test the relationship between post-sequencing MCCS scores and whether the sequencing identified a diagnosis (an ordinal variable defined as follows: 0=no diagnosis, 0.5=unconfirmed diagnosis, and 1=confirmed diagnosis). We report exact p-values and estimates of effect size for each statistical test. Because this is a small sample case series with low statistical power, we have interpreted the results of statistical tests using a more liberal p-value threshold of .10 [59]. We note, however, that these results are preliminary and should be viewed as observations that can inform future studies.

RESULTS

Participant characteristics

A total of 47 patients, parents, and physicians across 11 diagnostic odyssey cases participated in the study. On average, the length of relationship between physicians and patients was 38 months (SD = 39). Five of the 11 (45%) cases involved patients who were under 20 years of age. See Table 2 for a summary of demographic characteristics. See Supplemental Table 2 for a list of the genetic diagnoses for each of the N = 11 cases [60].

Physician-Patient Communication

Internal consistency for the MCCS was high with all versions having a Cronbach's $\alpha > .95$. In 6 of 11 cases (54%) the patient respondent rated the physician's communication of genome sequencing information as worse than communication of general medical information (pre-sequencing *Mdn* = 127,

post-sequencing *Mdn* = 123; $T = 9$, $p = .110$, $r = -.34$). In 9 of 11 cases (82%) physician self-ratings of communication of genome sequencing information were worse than the patient respondent ratings of the physician (patient respondent *Mdn* = 123, physician *Mdn* = 111; $T = 12$, $p = .062$, $r = -.40$). See Figure 1 for a graphical depiction of MCCS scores across respondents and time-points.

We also found that physician self-ratings of communication of genome sequencing information were positively associated with identification of a diagnosis ($r_s = .680$, $p = .021$), but diagnosis was not associated with patient respondent ratings of physicians' communication of genome sequencing information ($r_s = .018$, $p = .959$). See Figure 2 for a graphical depiction of MCCS scores as a function of diagnosis.

Decisional Regret

Internal consistency was high for all versions of the DRS (Cronbach's $\alpha > .85$). Post-sequencing DRS scores were comparable between patient respondents and physicians as both had a median DRS score of 0. However, while identification of a diagnosis was not associated with patient respondent DRS scores ($r_s = .105$, $p = .758$), diagnosis was negatively associated with physician DRS scores ($r_s = -.741$, $p = .009$) such that higher regret was associated with no diagnosis being found.

DISCUSSION

Findings from this small-sample preliminary case series suggest the hypothesis that even in diagnostic odyssey cases where genome sequencing has been shown to be clinically beneficial, physicians may not be well-equipped to communicate with patients and families about this type of information. Specifically, using a novel, multi-perspective approach to assess physician communication of patient genome sequencing information, we found that in most cases, patients and families rated the physician's communication of genomic information worse than general medical information. In addition, physicians tended to be even more self-critical, with their self-ratings being worse than how they were rated by patients and families. Consistent with published literature, this suggests that physicians would benefit from additional education and training in the use of WGS, including the best ways to convey WGS' strengths, limitations, and complexities to patients. This is particularly true as genomic applications continue to expand into clinical care [1], especially in the pediatrics field where genetic diseases constitute the majority of inpatient pediatric charges and admissions [36] and WGS has been shown to be a useful tool for pediatric clinical care [65]. One review of genomic medicine education initiatives across physician training level indicates some progress in electronic- and web-based resources, immersive experiences, and interprofessional and interdisciplinary learning [61]. Several of these initiatives have been studied and outcomes suggest they may result in increased confidence, motivation, and understanding in using genomic medicine among physicians [61]. Simulations

and standardized patients might also serve to improve knowledge and clinical skills [62-64].

We also found some evidence that identification of a diagnosis via WGS was related to how physicians viewed their own communication of the patient genome sequencing information, as well as whether or not the physician expressed regret regarding participation. In contrast, the identification of a diagnosis was not related to patient or family perceptions of communication or level of regret. The importance of diagnosis to physicians in our sample may reflect the well-known tendency of physicians to value medical tests and procedures with high clinical utility, whereas, many patients value information for information's sake (often termed "personal utility"). Therefore, patients and families in our study may have valued a finding of "no diagnosis" (more so than physicians) given that such an outcome provided another piece of information related to their diagnostic odyssey. With respect to decisional regret, specifically, we would emphasize that overall levels of regret among both patients and families, as well as physicians, were generally very low.

Limitations

A limitation of this study is the small sample size of 11 cases. Nevertheless, the rarity of diagnostic odyssey cases, coupled with the complexity of the sequencing protocol, make such cases both unusual and valuable from a research perspective. For instance, the study required biological parents to be available and willing to undergo sequencing

themselves. In addition, each patient participant had to have a “physician champion” available and willing to work with the research team and take responsibility for returning the sequencing results. Given physicians’ many time-constraints [66], recruiting a physician to participate could have been a logistical barrier that kept patients from being able to join the study. This particular criterion, however, allowed us to simulate a more realistic clinical encounter in which communication of genomic results might happen via a physician (versus a genetics specialist). Thus, each diagnostic odyssey case required the coordinated participation of several individuals in a complex protocol, which was executed over a period of several months. Nevertheless, we recognize the limitations of the small sample size and view the data in a hypothesis generative lens.

Patients involved in this study also represent a skewed demographic. They were mostly White, non-Hispanic, females from higher socioeconomic backgrounds with parents who attained high levels of education. Researchers have called attention to the demographic disparity in participatory genomic research [67, 68]. Numerous hypotheses have been proposed to explain the lower rates of participation in genomic research among minority groups including: (1) limited access to and awareness of genetic services [69-71]; (2) socioeconomic and sociocultural factors [69, 71, 72]; and (3) distrust in how genetic information might be used [69, 72]. It is of the utmost importance to consider and address systemic-level factors that deter

patients and families from underrepresented populations experiencing diagnostic odysseys from participating in genomic research.

Finally, the scales used in the assessment measures battery were modified from their original versions. Although measure modifications are known to affect reliability and validity of measures [73-75], our measure modifications were necessary in order to ensure that the assessments were specific to the genome sequencing scenario. Furthermore, this highlights the need for better assessment tools to understand physician, patient, and parent attitudes toward WGS [76] and other emerging technologies. Some evidence suggests that the information-verifying subscale of the MCCS may be the scale's most genuinely dyadic subscale [77], and future research on physician-patient dyads should consider its use.

One strength of this study is the multi-perspective approach towards understanding physician-patient communication of patient genome sequencing information. Unlike previous research that has taken a single perspective approach toward understanding genomic communication [4, 7-10, 12, 17, 18, 23-25, 44-47, 49, 50], our study considered communication of WGS results by including and comparing the perspectives of patients and parents, as well as physicians. The discordance in communication ratings between physician-patient dyads for the same return of genetic results session is noteworthy. Future research with larger samples that takes a multi-perspective approach is recommended. Furthermore, mixed methods approaches such as conducting follow-up qualitative interviews could benefit general

understanding of potential discrepancies in perceptions. For example, one of our cases (Case #3) was so discrepant that our study team elected to do the return of results a second time, but with a different physician (results not shown). Thus, the multi-perspective approach may be fruitful clinically as well, identifying cases where additional follow-up may be required.

Conclusion

Taken together, our findings suggest that further study of physician-patient communication of genomic information is warranted. Though limited inferences can be drawn from the small sample studied here, our results are consistent with other observations in the literature that physicians face challenges in communicating with their patients about this type of information. This work also points to the utility of multi-perspective study designs to assess and understand physician-patient communication of genome sequencing information. Most of the 850k practicing physicians in the U.S. trained prior to the completion of the Human Genome Project, and genomic medicine education is not consistently included in U.S. medical school curricula. As WGS becomes increasingly available in clinical settings, and physicians become more and more responsible for communicating genomic information, additional resources to support physicians in effective physician-patient communication is critical.

1

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Table 1 Primary measure respondent

Case Number	Primary Respondent
1	Mother
2	Patient
3	Mother
4	Patient
5	Patient
6	Patient
7	Patient
8	Mother
9	Mother
10	Mother
11	Patient

Table 2 Participant demographic characteristics

Demographic information, N = 11 cases	N (%)
Patient Gender	
-Female	8 (73)
-Male	3 (27)
Patient Age (years)	
-Under 12	3 (27)
-12 – 19	2 (18)
-20 – 27	1 (9)
-28 – 35	2 (18)
-36 – 43	2 (18)
-44+	1 (9)
Patient Race	
-White	11 (100)
Patient Ethnicity	
-Non-Hispanic	9 (82)
-Hispanic	2 (18)
Patient Education	
-None	2 (18)
-Some grade school	1 (9)
-Some high school	1 (9)
-College (current)	1 (9)
-College degree	6 (55)
Mother’s Education	
-High School Degree	2 (18)
-College Degree	7 (64)
-Graduate Degree	1 (9)
-Not Reported	1 (9)
Father’s Education	
-College Degree	7 (64)
-Graduate Degree	3 (27)
-Not Reported	1 (9)
Income	
-$25,000-$49,999	1 (9)
-$50,000-$74,999	1 (9)
-$150,000-$199,999	1 (9)
-$200,000-$249,999	2 (18)
-$250,000-$299,999	1 (9)
-Not Reported	5 (45)
Physician Medical Specialty	
-Oncology/Hematology	3 (27)
-Genetics	2 (18)
-Allergy/Immunology	1 (9)
-Cardiology	1 (9)
-Gastroenterology	1 (9)

-General Surgery -Neurology -Pediatrics	1 (9) 1 (9) 1 (9)
Diagnosis Status -No diagnosis found -Probable diagnosis -Confirmed diagnosis	 3 (27) 6 (54) 2 (18)
Physician-Patient Length of Relationship (months) -Less than 12 months -12-60 months -61-120 months	 3 (27) 5 (45) 3 (27)

1
2

Supplemental Table 1 Assessment measures battery

Assessment Measure	Patient		Parents		Physician	
	Pre	Post	Pre	Post	Pre	Post
World Health Organization Quality of Life [78] (Modified)	X					
Factual Genetic Knowledge Assessment [79, 80] (Modified)	X					
Whole Genome Sequencing Questionnaire [81] (Modified)	X	X	X	X	X	X
Attitudes Toward Secondary Findings*	X	X	X	X	X	X
Personal Involvement Inventory [82]	X	X	X	X	X	X
Medical Communication Competence Scale [56] (Modified)	X	X	X	X		X
Decision Regret Scale [57] (Modified)		X		X		X
Genome Sequencing Utility/Understanding *		X		X		X
Impact of Events Scale [83, 84] (Modified)		X				
Diffusion of Innovations [85] (Modified)					X	

* Measure was developed in-house for the purposes of this study

Supplemental Table 2 Genetic diagnoses of enrolled cases

Case	Phenotype	Outcome	Plausible diagnosis	Functional confirmation
1	Complex movement disorder	*ADCY5* de novo gain-of-function mutation	Yes	Yes
2	Lymphoproliferative disorder	Plausible inherited candidate causative mutations in *GIMAP8*	Yes	Not performed
3	Inflammatory bowel disease	Plausible inherited candidate causative mutations in *MST1R*	Yes	Not performed
4	Fibromyxoid sarcoma	No cause identified	No	N/A
5	Immunodeficiency	No cause identified	No	N/A
6	Lipomatosis and vasculitis	Plausible inherited candidate causative mutations in *NRIP1*	Yes	Not performed
7	Skin and neurological disorder	Potential but unconfirmed diagnosis of a subtype of epidermolysis bullosa due to inherited mutations in *DST1*	Yes	Ongoing
8	Developmental delay	No cause identified	No	N/A
9	Epileptic encephalopathy	*KCNB1* de novo missense mutation and confirmed KV2.1 dysfunction	Yes	Yes
10	Developmental delay	Plausible inherited candidate causative mutations in *SYNPO*	Yes	N/A
11	Familial coronary artery disease	Plausible inherited candidate causative mutation in *TDG*	Yes	Ongoing

Figure 1. Medical Communication Competence Scale (MCCS) Scores for patient respondents and physicians.

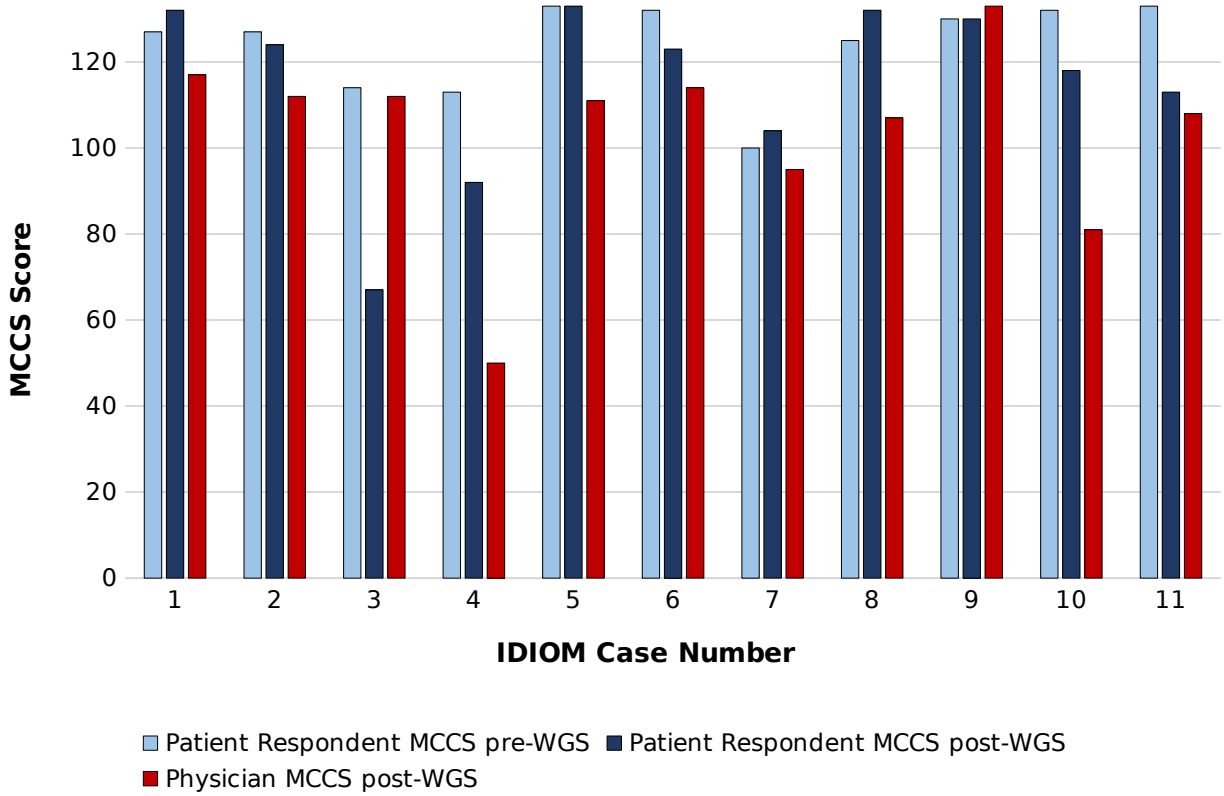
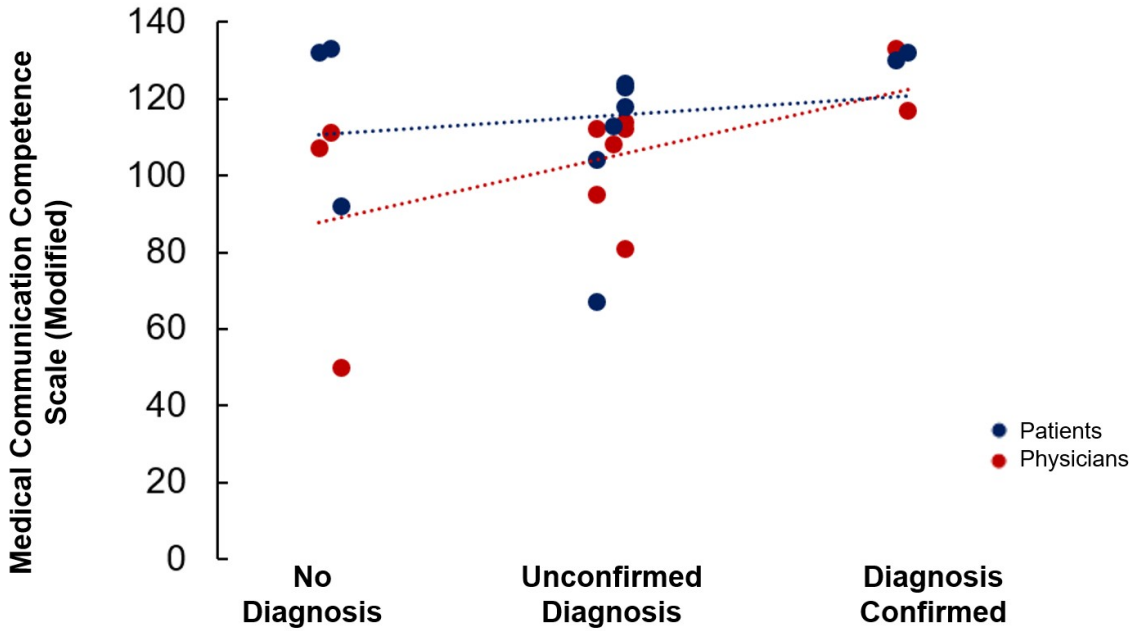


Figure 2. Relationship between receipt of a diagnosis and post-sequencing Medical Communication Competence Scale (MCCS) Scores for patient respondents and physicians.



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APPENDIX A – MEDICAL COMMUNICATION COMPETENCE SCALES (MODIFIED)

Patient Pre-sequencing Medical Communication Competence Scale (Modified)

Please think about the **communications you have had with your physician about your medical problems.**

Please show how strongly you agree or disagree with these statements by **checking the box that best fits your views about your decision**.

		Strongly Disagree	Disagree	Slightly Disagree	Not Sure	Slightly Agree	Agree	Strongly Agree
1.	My physician generally seems knowledgeable							
2.	My physician seems knowledgeable about my medical problems							
MY PHYSICIAN EXPLAINS THE FOLLOWING TO MY SATISFACTION:								
3.	What my medical problems are							
4.	The possible causes of my medical problems							
5.	What I may be able to do to get better							
6.	The benefits and disadvantages of treatment choices, if any							
7.	The purpose of any tests that are needed							
8.	How any prescribed medicines may help my medical problems							
9.	The long-term consequences of my medical problems							
MY PHYSICIAN DOES A GOOD JOB OF:								
10.	Reviewing or repeating important information							

11.	Making sure I understand his/her explanations							
12.	Making sure I understand his/her directions							
13.	Using language I can understand							
14.	Checking his/her understanding of what I say							
15.	Encouraging me to ask questions							
Please indicate how strongly you agree or disagree with these statements:								
16.	My physician seems comfortable discussing my medical problems							
17.	My physician seems to understand my medical problems him/herself							
18.	My physician spends enough time explaining/discussing my medical problems							
19.	Overall, I am satisfied with my physician’s communication regarding my medical problems							

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Parent Pre-sequencing Medical Communication Competence Scale (Modified)

Please think about the **communications you have had with your child's physician about your child's medical problems.**

Please show how strongly you agree or disagree with these statements by **checking the box that best fits your views about your decision**.

		Strongly Disagree	Disagree	Slightly Disagree	Not Sure	Slightly Agree	Agree	Strongly Agree
1.	My child's physician generally seems knowledgeable							
2.	My child's physician seems knowledgeable about my child's medical problems							
MY CHILD'S PHYSICIAN EXPLAINS THE FOLLOWING TO MY SATISFACTION:								
3.	What my child's medical problems are							
4.	The possible causes of my child's medical problems							
5.	What my child may be able to do to get better							
6.	The benefits and disadvantages of treatment choices, if any							
7.	The purpose of any tests that are needed							
8.	How any prescribed medicines may help my child's medical problems							
9.	The long-term consequences of my child's medical problems							
MY CHILD'S PHYSICIAN DOES A GOOD JOB OF:								
10.	Reviewing or repeating important information							
11.	Making sure I understand his/her explanations							

12.	Making sure I understand his/her directions							
13.	Using language I can understand							
14.	Checking his/her understanding of what I say							
15.	Encouraging me to ask questions							
Please indicate how strongly you agree or disagree with these statements:								
16.	My physician seems comfortable discussing my child's medical problems							
17.	My physician seems to understand my child's medical problems him/herself							
18.	My physician spends enough time explaining/discussing my child's medical problems							
19.	Overall, I am satisfied with how my child's physician communicates regarding my child's medical problems							

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Patient Post-sequencing Medical Communication Competence Scale (Modified)

Please think about the **communications you've had with your physician about your whole genome sequencing test results**.

Please show how strongly you agree or disagree with these statements by **checking the box that best fits your views about your decision**.

		Strongly Disagree	Disagree	Slightly Disagree	Not Sure	Slightly Agree	Agree	Strongly Agree
1.	My physician seemed knowledgeable about genetics in general							
2.	My physician seemed knowledgeable about whole genome sequencing							
MY PHYSICIAN EXPLAINED THE FOLLOWING TO MY SATISFACTION:								
3.	What my whole genome sequencing results were							
4.	The relationship between my medical problem and my whole genome sequencing results, if any							
5.	How my whole genome sequencing results may or may not help me get better							
6.	The benefits and disadvantages of treatment choices, if any, based on my whole genome sequencing results							
7.	The purpose of any additional tests that were needed as follow-up to my whole genome sequencing							
8.	How any prescribed medicines based on the results of my whole genome sequencing may help my medical problem							
9.	The long-term consequences of my whole genome sequencing results							

MY PHYSICIAN DID A GOOD JOB OF:								
10.	Reviewing or repeating important information about my whole genome sequencing results							
11.	Making sure I understood his/her explanations of my whole genome sequencing results							
12.	Making sure I understood his/her directions related to medical follow-up from my whole genome sequencing results, if any							
13.	Using language I could understand related to my whole genome sequencing results							
14.	Checking his/her understanding of what I said about my whole genome sequencing results							
15.	Encouraging me to ask questions about my whole genome sequencing results							
Please indicate how strongly you agree or disagree with these statements:								
16.	My physician seemed comfortable discussing my whole genome sequencing results							
17.	My physician seemed to understand the results of my whole genome sequencing results him/herself							
18.	My physician spent enough time explaining/discussing my whole genome sequencing results							
19.	Overall, I am satisfied with my physician’s delivery of my whole genome sequencing test results							

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Parent Post-sequencing Medical Communication Competence Scale (Modified)

Please think about the **communications you've had with your child's physician about your child's whole genome sequencing test results**.

Please show how strongly you agree or disagree with these statements by **checking the box that best fits your views about your decision**.

		Strongly Disagree	Disagree	Slightly Disagree	Not Sure	Slightly Agree	Agree	Strongly Agree
1.	My child's physician seemed knowledgeable about genetics in general							
2.	My child's physician seemed knowledgeable about whole genome sequencing							
MY CHILD'S PHYSICIAN EXPLAINED THE FOLLOWING TO MY SATISFACTION:								
3.	What my child's whole genome sequencing results were							
4.	The relationship between my child's medical problem and my child's whole genome sequencing results, if any							
5.	How my child's whole genome sequencing results may or may not help him/her get better							
6.	The benefits and disadvantages of treatment choices, if any, based on my child's whole genome sequencing results							
7.	The purpose of any additional tests that were needed as follow-up to my child's whole genome sequencing							
8.	How any prescribed medicines based on the results of my child's whole genome sequencing may help his/her medical problem							
9.	The long-term consequences of my child's whole genome sequencing results							

MY CHILD'S PHYSICIAN DID A GOOD JOB OF:								
10.	Reviewing or repeating important information about my child's whole genome sequencing results							
11.	Making sure I understood his/her explanations of my child's whole genome sequencing results							
12.	Making sure I understood his/her directions related to medical follow-up from my child's whole genome sequencing results, if any							
13.	Using language I could understand related to my child's whole genome sequencing results							
14.	Checking his/her understanding of what I said about my child's whole genome sequencing results							
15.	Encouraging me to ask questions about my child's whole genome sequencing results							
Please indicate how strongly you agree or disagree with these statements:								
16.	My child's physician seemed comfortable discussing my child's whole genome sequencing results							
17.	My child's physician seemed to understand the results of my child's whole genome sequencing results him/herself							
18.	My child's physician spent enough time explaining/discussing my child's whole genome sequencing results							
19.	Overall, I am satisfied with how my child's physician delivered my child's whole genome sequencing test results							

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Physician Post-sequencing Medical Communication Competence Scale (Modified)

Please think about the **communications you've had with your patient about his/her whole genome sequencing test results**.

Please show how strongly you agree or disagree with these statements by **checking the box that best fits your views about your decision**.

		Strongly Disagree	Disagree	Slightly Disagree	Not Sure	Slightly Agree	Agree	Strongly Agree
I PROVIDED GOOD EXPLANATIONS OF THE FOLLOWING TO THE PATIENT:								
1.	What my patient's whole genome sequencing results were							
2.	The relationship between my patient's medical problem and his/her whole genome sequencing results, if any							
3.	How my patient's whole genome sequencing results may or may not help improve his/her health							
4.	The benefits and disadvantages of treatment choices, if any, based on his/her whole genome sequencing results							
5.	The purpose of any additional tests that were needed as follow-up to his/her whole genome sequencing							
6.	How any prescribed medicines based on the results of my patient's whole genome sequencing may help his/her medical problem							
7.	The long-term consequences of my patient's whole genome sequencing results							
I DID A GOOD JOB OF:								
8.	Reviewing or repeating important information about my patient's whole genome sequencing results							

9.	Making sure my patient understood my explanations of his/her whole genome sequencing results							
10.	Making sure my patient understood my directions related to medical follow-up from his/her whole genome sequencing results, if any							
11.	Using language my patient could understand related to his/her whole genome sequencing results							
12.	Checking my understanding of what my patient said about his/her whole genome sequencing results							
13.	Encouraging my patient to ask questions about his/her whole genome sequencing results							
Please indicate how strongly you agree or disagree with these statements:								
14.	I was comfortable discussing my patient's whole genome sequencing results							
15.	I understand my patient's whole genome sequencing results myself							
16.	I spent enough time explaining/discussing my patient's whole genome sequencing results							
17.	Overall, I am satisfied with the way I communicated my patient's whole genome sequencing test results to him/her							
18.	I feel I am knowledgeable enough about genetics to discuss my patient's whole genome sequencing results with him/her							
19.	I feel I am knowledgeable enough about whole genome sequencing to discuss my patient's whole genome sequencing results with him/her							

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APPENDIX B – DECISION REGRET SCALES (MODIFIED)

Patient Decision Regret Scale (Modified)

Please reflect on the decision to undergo **whole genome sequencing**. Please show how strongly you agree or disagree with these statements by **checking the box that best fits your views about your decision**.

		Strongly disagree	**Disagree**	**Neither agree nor disagree**	**Agree**	**Strongly agree**
1.	It was the right decision					
2.	I regret the choice that was made					
3.	I would go for the same choice if I had to do it over again					
4.	The choice did me a lot of harm					
5.	The decision was a wise one					

Parent Decision Regret Scale (Modified)

Please reflect on the decision to undergo **whole genome sequencing**. Please show how strongly you agree or disagree with these statements by **checking the box that best fits your views about your decision**.

		Strongly disagree	**Disagree**	**Neither agree nor disagree**	**Agree**	**Strongly agree**
1.	It was the right decision					
2.	I regret the choice that was made					
3.	I would go for the same choice if I had to do it over again					
4.	The choice did me a lot of harm					
5.	The decision was a wise one					

Physician Decision Regret Scale (Modified)

Please reflect on the decision to act as the “physician champion” for your patient who underwent **whole genome sequencing**. Please show how strongly you agree or disagree with these statements by **checking the box that best fits your views about your decision**.

		Strongly disagree	Disagree	Neither agree nor disagree	Agree	Strongly agree
1.	It was the right decision					
2.	I regret the choice that was made					
3.	I would go for the same choice if I had to do it over again					
4.	The choice did me and/or my patient a lot of harm					
5.	The decision was a wise one					