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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

1 MCCS: Medical Communication Competence Scale

2 U.S.: United States

3 WGS: Whole genome sequencing

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

7
8 This multi-perspective study aims to gain further insight into physician
9 communication of patient genome sequencing information.

10
11
12 **What's Known on This Subject** (max 40 words, in paragraph style)

13
14 Research has largely taken a single-perspective (e.g., physician perspective
15 or patient perspective) approach towards understanding genomic results-
16 related communication. Moreover, previous findings suggest that as a group,
17 physicians face challenges in effectively communicating patient genome
18 sequencing information.

19
20
21 **What This Study Adds** (max 40 words, in paragraph style)

22
23 This study describes a multi-perspective research approach to study
24 physician communication of patient genome sequencing information in
25 diagnostic odyssey cases. Findings suggest that physician communication of
26 patient genome sequencing information is suboptimal compared to
27 communication of general patient medical information.
28

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

1 INTRODUCTION

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

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

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

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

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

hypotheses for future research on novel clinical phenomena⁵⁴. 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 WGS⁵⁵, and prior work in the context of IDIOM has also explored patient perspectives on participation in the study using qualitative methods⁴⁵.

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 elsewhere⁵⁵. 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

1 declarative statements⁵⁶. The MCCS has shown adequate internal
2 consistency (Cronbach's $\alpha = .75-.90$)⁵⁶. Statements on the MCCS were
3 written from the perspective of the respondent such that a patient received a
4 version written from a patient's perspective (e.g., *My physician...*) while a
5 parent received a version written from the perspective of a parent (e.g., *My*
6 *child's physician...*). See Appendix A for a copy of the modified MCCS that
7 was used. Sample statements include, *My physician seems knowledgeable*
8 *about my medical problems* and *My child's physician does a good job of*
9 *reviewing or repeating important information*. At the pre-sequencing time-
10 point, the patient respondent was asked to rate their physician's
11 communication of general medical information. Only patient respondents
12 completed the MCCS at the pre-sequencing time-point (Supplemental Table
13 1).

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

22 *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 statements⁵⁷. The scale has shown adequate internal consistency (Cronbach's $\alpha = .81-.92$)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

1 addition, we used a Spearman rank correlation to test the relationship
2 between post-sequencing MCCS scores and whether the sequencing
3 identified a diagnosis (an ordinal variable defined as follows: 0=no diagnosis,
4 0.5=unconfirmed diagnosis, and 1=confirmed diagnosis). We report exact p-
5 values and estimates of effect size for each statistical test. Because this is a
6 small sample case series with low statistical power, we have interpreted the
7 results of statistical tests using a more liberal p-value threshold of .10⁵⁹. We
8 note, however, that these results are preliminary and should be viewed as
9 observations that can inform future studies.

11 **RESULTS**

12 **Participant characteristics**

13 A total of 47 patients, parents, and physicians across 11 diagnostic
14 odyssey cases participated in the study. On average, the length of
15 relationship between physicians and patients was 38 months (SD = 39). Five
16 of the 11 (45%) cases involved patients who were under 20 years of age. See
17 Table 2 for a summary of demographic characteristics. See Supplemental
18 Table 2 for a list of the genetic diagnoses for each of the N = 11 cases⁶⁰.

19 **Physician-Patient Communication**

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

1 post-sequencing $Mdn = 123$; $T = 9$, $p = .110$, $r = -.34$). In 9 of 11 cases
2 (82%) physician self-ratings of communication of genome sequencing
3 information were worse than the patient respondent ratings of the physician
4 (patient respondent $Mdn = 123$, physician $Mdn = 111$; $T = 12$, $p = .062$, $r =$
5 $-.40$). See Figure 1 for a graphical depiction of MCCS scores across
6 respondents and time-points.

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

13 **Decisional Regret**

14 Internal consistency was high for all versions of the DRS (Cronbach's α
15 $> .85$). Post-sequencing DRS scores were comparable between patient
16 respondents and physicians as both had a median DRS score of 0. However,
17 while identification of a diagnosis was not associated with patient respondent
18 DRS scores ($r_s = .105$, $p = .758$), diagnosis was negatively associated with
19 physician DRS scores ($r_s = -.741$, $p = .009$) such that higher regret was
20 associated with no diagnosis being found.

22 **DISCUSSION**

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

1 and standardized patients might also serve to improve knowledge and
2 clinical skills ⁶²⁻⁶⁴.

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

18 **Limitations**

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

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

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

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

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

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

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

6 **Conclusion**

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

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1
2
3

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

4
5

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

1
2

1 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	

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

3

1

2 Supplemental Table 2 Genetic diagnoses of enrolled cases

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

4

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

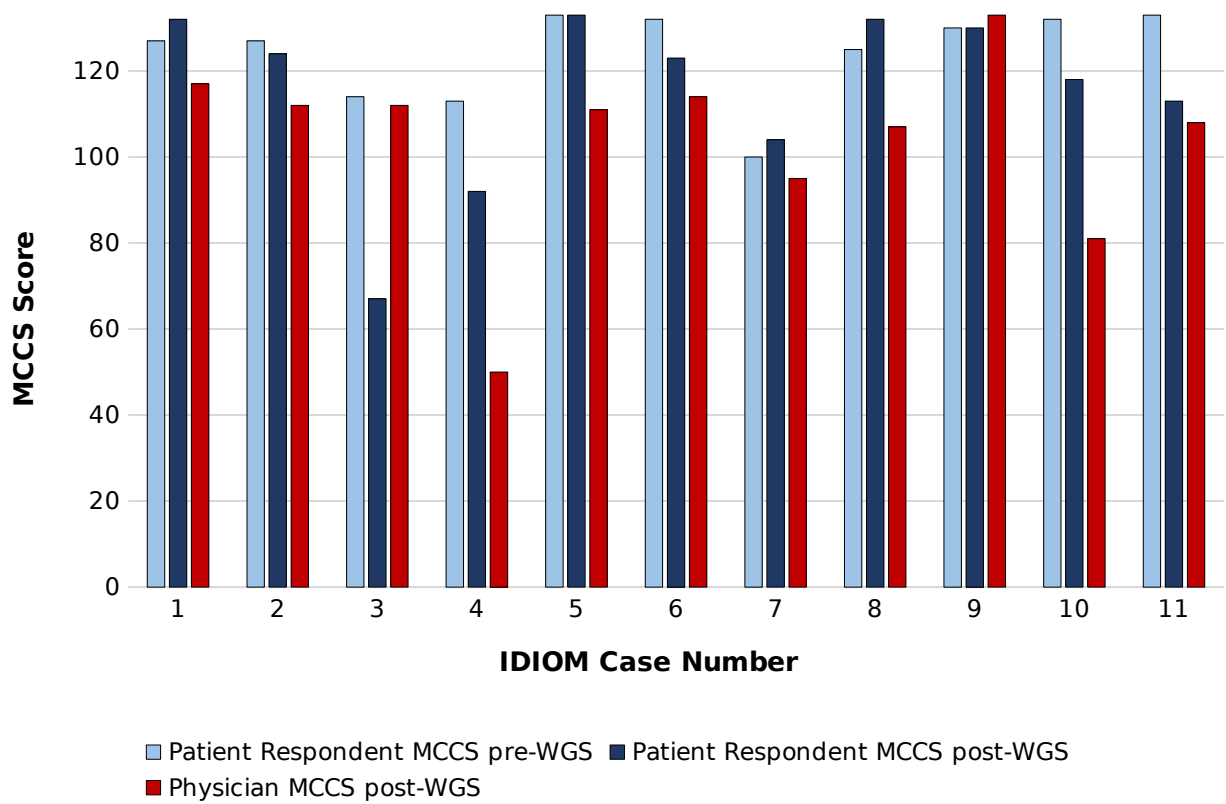
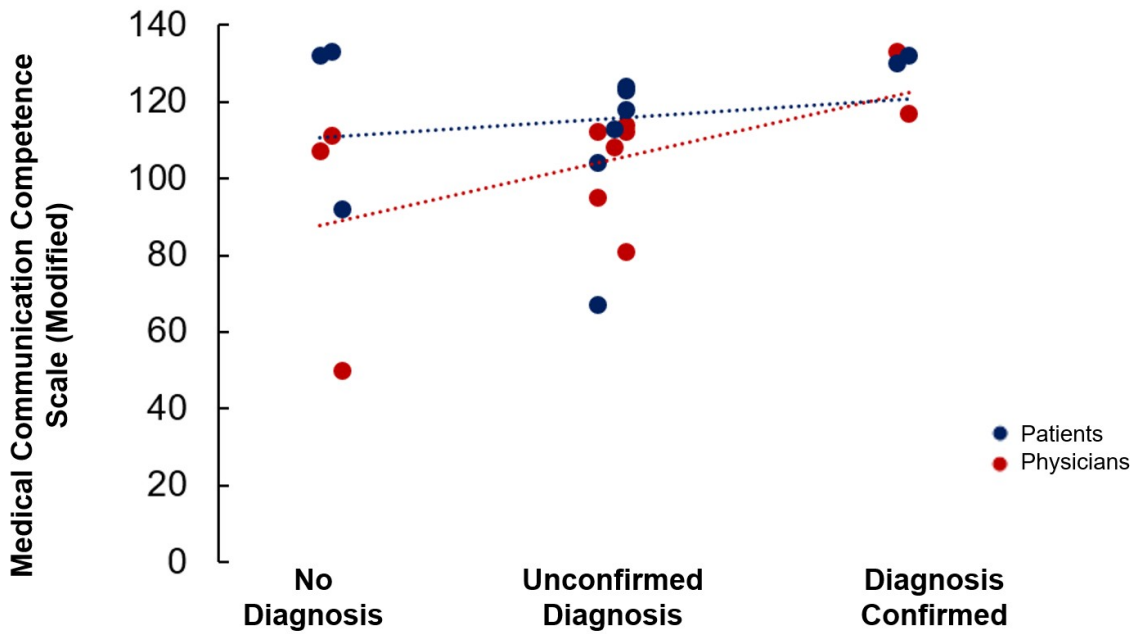


Figure 2. Relationship between receipt of a diagnosis and post-sequencing Medical Communication Competence Scale (MCCS) Scores for patient respondents and physicians.



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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.**

		Strongl y Disagr ee	Disagr ee	Slightl y Disagr ee	Not Sure	Slightl y Agree	Agree	Strongl y 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.**

		Strongl y Disagr ee	Disagr ee	Slightl y Disagr ee	Not Sure	Slightl y Agree	Agree	Strongl y 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.**

		Strongl y Disagr ee	Disagr ee	Slightl y Disagr ee	Not Sure	Slightl y Agree	Agree	Strongl y 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.**

		Strongl y Disagr ee	Disagr ee	Slightl y Disagr ee	Not Sure	Slightl y Agree	Agree	Strongl y 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.**

		Strongl y Disagr ee	Disagr ee	Slightl y Disagr ee	Not Sure	Slightl y Agree	Agree	Strongl y 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					

1 **Parent Decision Regret Scale (Modified)**

2
3 Please reflect on the decision to undergo **whole genome sequencing**. Please show
4 how strongly you agree or disagree with these statements by **checking the box**
5 **that best fits your views about your decision.**

6

		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					

1 **Physician Decision Regret Scale (Modified)**

2
3 Please reflect on the decision to act as the “physician champion” for your patient who
4 underwent **whole genome sequencing**. Please show how strongly you agree or
5 disagree with these statements by **checking the box that best fits your views**
6 **about your decision.**

7

		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					

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