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Parent-Reported Clinical Utility of Pediatric Genomic Sequencing

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Abstract

Background and Objectives: Genomic sequencing (GS) is increasingly used for diagnostic evaluation, yet implications for follow-up care are not well understood. We assessed clinicians'

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Contributors' Statement

Dr. Smith conceptualized and designed the study, carried out the initial analyses, drafted the initial manuscript, and reviewed and revised the manuscript. Drs. Ferket, Gelb, Hindorff, Norton, Ferar, Sahin-Hodoglugil, Slavotinek, Lich, Berg, and Russell conceptualized and designed the study and reviewed and revised the manuscript. All authors revised and approved the final manuscript as submitted and agree to be accountable for all aspects of the work.

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recommendations following GS and whether parents followed up on them, as well as actions parents initiated themselves in response to learning their child's GS results.

Methods: We surveyed parents of children who received GS through the Clinical Sequencing Evidence Generating Research (CSER) consortium approximately 5–7 months after return of results. We compared the proportion of parents who reported discussing their child's result with a clinician, clinicians' recommendations, and parents' follow-up actions between parents of children who received positive, inconclusive, and negative GS results using chi-squared tests.

Results: A total of 1,188 respondents completed survey measures on recommended medical actions (RMA, n=1,187) and/or parent-initiated actions (PIA, n=913), 2018 – 2022. Most parents who completed RMA questions (n=833, 70.3%) reported having discussed their child's GS results with their child's clinicians. Clinicians made recommendations to change current care for patients with positive GS results (n=79, 39.1%) more frequently than for those with inconclusive (n=31, 12.4%) or negative results (n=44, 11.9%; p<0.001). Many parents discussed (n=152 completed, n=135 planned) implications of GS results for future pregnancies with a clinician. Aside from clinical recommendations, 13.0% (n=119) of parents initiated changes to their child's health or lifestyle.

Conclusions: In diverse pediatric clinical contexts, GS results can lead to recommendations for follow up care, but they likely do not prompt large increases in the quantity of care received or lifestyle and behavior changes.

Article Summary

Surveys of parents of pediatric patients who received genomic sequencing in the CSER consortium regarding clinicians' recommendations and parent-initiated actions.

Introduction

Genomic sequencing (GS) is increasingly used in pediatric clinical diagnostic evaluation. While many studies have demonstrated the diagnostic utility of GS for newborns, infants, and children in both inpatient and outpatient settings,[1–7] there is relatively little evidence on the effect of GS results on clinical decision making and downstream clinical care delivered in diverse pediatric settings.[8,9] Knowledge of GS results, whether or not they lead to a new molecular diagnosis, has the potential to inform prognostication, clinical care trajectories, and family decision making.[10,11] However, patients and their families may also face uncertainty regarding what the result means for their clinical care and other aspects of their life.

When GS results do have direct implications for the child's care, families may encounter barriers to accessing recommended care for many reasons that remain understudied.[12] Survey data from parents of pediatric patients is valuable to understanding both clinical and personal impacts of GS on families. Parents are an important source of information regarding actions that their child's clinicians recommended and any follow-up care that their child received because of GS results. Additionally, parents may initiate changes to aspects of family life aside from clinical care, such as lifestyle or behavioral modifications, changes to employment, or uptake of new insurance policies. Large-scale efforts to systematically

collect evidence on actions following the return of GS results that occur both inside and outside of clinical care can help better characterize the effect of GS on families.

The Clinical Sequencing Evidence-Generating Research (CSER) consortium provides a unique opportunity to examine clinicians' recommendations, follow-up on downstream clinical care attributable to GS, and parent-initiated actions for patients with diverse clinical indications. In its second phase of funding, the CSER consortium included six extramural clinical sequencing projects and one National Human Genome Research Institute intramural project, each of which had the goal of enrolling at least 60% of participants who were medically underserved or historically underrepresented in genomics research, as well as a data coordinating center.[13] A major goal of this NIH-funded consortium was to generate evidence on the personal and clinical utility of GS through surveys of participants and clinicians.[14] Five CSER projects enrolled pediatric patient-participants, and parental outcomes were assessed across a range of domains including psychosocial outcomes, perceived utility, understanding and information seeking, and actions attributable to GS. We report findings from portions of the harmonized surveys that assessed attributable actions.

We describe clinical care recommendations, follow-up care received, and actions parents self-initiated after receiving their child's GS results through CSER consortium pediatric projects. Parental survey data is presented according to whether the child received a positive, negative, or inconclusive GS result. Our findings provide insight into how parents of children who undergo clinically indicated GS use the results inside and outside of clinical care settings.

Methods

CSER Consortium Projects

Five projects within the CSER consortium enrolled patient-participants less than 18 years of age in multiple geographic regions across the US (Table 1). Patient cohorts included pediatric cancer patients (KidsCanSeq), critically ill newborns (SouthSeq), pediatric and prenatal patients with developmental disorders or structural anomalies (P^3EGS), and pediatric patients with various undiagnosed disorders, including neurodevelopmental disorders (NCGENES 2) and neurologic, immunologic, and cardiac disorders (NYCKidSeq). We focused on pediatric and prenatal projects, rather than projects that enrolled adults, so that all survey data were parent-reported.

Survey instruments

The CSER consortium harmonized survey measures across projects that were designed to assess the multi-dimensional impacts of GS.[14] The final survey was scheduled for 5–7 months after GS results were returned to families. The Clinical Utility, Health Economics and Policy (CUHEP) Working Group, comprised of geneticists, medical specialists, economists, health services researchers, health policy experts, and others, developed survey questions to assess downstream medical recommendations and actions.

The CUHEP portion of the survey consisted of two domains: 1) recommended medical actions (RMA), the actions that their child's clinician(s) had recommended in response to their child's GS result; and 2) parent-initiated actions (PIA), the actions that parents initiated on their own which the clinician did not specifically recommend based on their child's GS result. Both domains included close-ended and open-ended questions. Multiple selections were possible for clinical recommendations and parent-initiated actions. The specific instruments used in this analysis are provided as Supplementary Material.

Survey administration was performed by each project's research team after project-specific institutional review board approval. Surveys were available in English and Spanish. Although harmonized across the consortium, project-specific survey adaptations were made as needed, and some projects administered surveys to only a subset of participants (Table 1). Two projects did not administer PIA questions because they were deemed by project leadership to be inappropriate for the clinical context (i.e., children with cancer and critically ill newborns). Similarly, RMA questions regarding referrals for mental health support, therapeutic services, and lifestyle changes were not administered to parents of critically ill newborns, as asking parents these questions while their child was ill might have been perceived as insensitive.

Genomic sequencing and variant interpretation

Genome or exome sequencing, either alone or in combination with tumor sequencing, was performed for each patient-participant. Each CSER team conducted independent sequence analysis and variant classification for participants enrolled in their study according to their own internal standards, consistent with guidelines published by the American College of Medical Genetics and Genomics and the Association for Molecular Pathology.[15] In addition, the consortium developed a framework for case-level interpretation of genetic findings, to contextualize the variant findings with the clinical presentation. Case-level GS results were interpreted by each project as positive, inconclusive, or negative. Briefly, positive results represent genetic and phenotypic findings that are consistent with a monogenic disease and its mode of inheritance, while negative results represent no reportable genetic findings. In this framework there are several types of inconclusive results in which there may be uncertainty regarding variant pathogenicity, phenotypic fit, allelic requirement, and/or parental inheritance. Full details of the sequencing results will be reported individually by each research group and collectively in forthcoming consortium manuscripts. Some patients had more than one variant identified; we grouped them according to the most significant result. All sites uploaded their GS results to a central REDCap[16] data repository managed by the consortium Data Coordinating Center.

Analytic data set

We constructed the analytic data set using survey results uploaded from the pediatric CSER projects into the central repository through July 2022, approximately one year after enrollment ended. We included survey responses from parents if their child had exome or genome sequencing results available, the results were returned to the parent, and the parent completed RMA and/or PIA questions. We matched survey responses to GS results using

a consortium-assigned patient identification number and analyzed all available data without imputation of any missing responses.

We used descriptive statistics to summarize survey responses and chi-squared tests to assess whether survey responses were dependent upon case-level GS result categories. Results from unstratified analyses are reported, as they were consistent with analyses stratified by project. Results with $p<0.05$ were considered significant. We coded free-text responses to open-ended questions and grouped similar codes to develop themes. Analysis was conducted in Stata 17 (StataCorp, College Station, TX) and Excel (Microsoft Corporation).

Results

A total of 2,481 parents with CSER identification numbers in both survey and genetic data and who met inclusion criteria were included in the analytic data set. Of those respondents, 47.9% (n=1,188) completed one or both CUHEP measures (RMA=1,187; PIA=913). Surveys were completed between July 2018 and May 2022, and 81.8% (n=972) were completed on or after April 1, 2020, during the COVID-19 pandemic which disrupted access to some health care services. The respondents' children had undergone various forms of testing, including exome or genome sequencing (Table 2). On average, respondents who completed RMA and/or PIA questions submitted their responses approximately 31 weeks after their child's GS results were returned.

Among respondents who completed the CUHEP measures, the patient-participant was still alive at the time of the survey in 87.6% (n=1,038) of cases, as compared to 83.0% (n=1,068) among respondents who did not complete the measures (p=0.001). Most respondents were parents of pediatric patients (85.0%), 15.0% (n=178) of respondents were pregnant patients, and 13.8% (n=163) of surveys were completed in Spanish (Table 2). Using the CSER Underserved Framework (described elsewhere), 53.1% (n=516) of respondents who completed the CUHEP measures were considered to be at risk for experiencing barriers to accessing clinical care, as compared to 69.3% (n=591) of those who did not complete the measures ($p<0.001$, Table 2).

Recommended Medical Actions

Among respondents who completed RMA questions (n=1,187), 70.3% (n=833/1,185) reported having discussed their child's genetic test results with their child's doctors or health care providers (henceforth referred to as "clinicians") other than those directly involved in the research protocol who returned the GS results to the family. The proportion of respondents who reported talking with their child's clinician was highest among those whose child received a positive GS result (84.3%), compared with those who received an inconclusive (63.1%) or negative result (69.5%; $p<0.001$). Additionally, 20.0% (n=237) of respondents reported that they had not yet discussed the results with their child's doctor but planned to do so (Table 3). Respondents who did not plan to discuss the results with their child's clinicians (n=115, 9.7%) indicated in free-text responses (n=94) that they felt the information did not seem relevant or important, that it could not improve their child's care, or that the doctors already knew the result or could see it in the electronic medical record (EMR).

Among the 833 respondents who discussed the results with their child's clinicians, respondents reported sharing results with primary care providers/pediatricians (n=377, 45.3%), neurologists (n=285, 34.2%), oncologists (n=241, 28.9%), cardiologists (n=23, 2.8%), and other specialists (n=179, 21.5%). Other specialists, as indicated in free-text responses (n=59), included endocrinologists, geneticists and genetic counselors, fertility specialists, hematologists, and gastroenterologists. Among respondents who discussed results with their child's clinician, 39.1% (n=79/202) of respondents whose child had a positive GS result reported that their clinician made recommendations to change current care, compared to 12.4% (n=31/250) of those with an inconclusive finding and 11.9% (n=44/369) of those with a negative result ($p<0.001$, Table 3).

The care changes that clinicians recommended are detailed in Table 4. Among respondents who reported that one or more clinical recommendations were made (n=154), 52.6% (n=81) did not select any of the pre-specified response options for types of clinical recommendations. Of those who did report at least one specific clinical recommendation, 40.3% (n=62) reported having already followed the recommendation(s), 5.8% (n=9) reported that they planned to follow the recommendation(s), and 1.3% (n=2) reported that they did not plan to follow the recommendation(s).

Clinicians sometimes made counseling recommendations regarding future pregnancies. After receiving their child's GS results, 26.6% (n=49/184) of respondents whose child received a positive result reported receiving counseling from an OB/GYN, reproductive genetic counselor, or primary care provider to discuss how their child's diagnosis might affect future pregnancies, while 10.0% (n=21/211) of those with an inconclusive finding and 23.8% (n=82/344) of those with a negative finding reported receiving counseling ($p<0.001$). Another 15.2%, 19.4%, and 19.2% of respondents whose child received a positive, inconclusive, or negative result, respectively, reported that they had not yet had discussions about what their child's results might mean for future pregnancies, but that they planned to do so. Approximately one-third of parents whose child received a positive finding (32.6%) or a negative finding (39.0%), and approximately one-half of parents whose child received an inconclusive finding (52.6%), reported that a discussion about how the diagnosis might affect future pregnancies was not applicable, while 448 respondents left the question blank. In free-text responses (n=123), respondents who indicated that counseling for future pregnancies was not applicable explained that they did not plan to have additional children, that their child's condition was not inherited or there was a very small recurrence risk, that the findings would not impact their decision whether to have additional children, or that it did not seem relevant or pressing to discuss the results.

Parent-Initiated Actions

A total of 913 respondents completed the PIA measure, of whom 13.0% (n=119/912) reported that they made changes to their child's health care or lifestyle based on GS results (positive n=33; inconclusive n=39; negative n=47) aside from any medical recommendations made by a clinician. The most frequently reported type of change overall was a change in diet (n=73, 8.0%) followed by a change in exercise (n=35, 3.8%), followed by starting vitamins and supplements (n=28, 3.1%). Thirty-five respondents whose children received

positive (n=11), inconclusive (n=14), or negative (n=10) GS results provided free-text responses noting other changes that they had made, including being more acutely aware of their child's care or sensitivities to specific activities or chemicals that could trigger symptoms, being more understanding or patient with their child, changing therapies (speech, occupational, behavioral), increasing the amount of sleep their child gets, and leaving or changing a job.

Four respondents reported changing their child's insurance based on their child's GS results. Two respondents bought new or higher coverage life insurance; no new or more generous disability or long-term care insurance purchases were reported. Thirty-eight (4.2%) respondents reported making some other lifestyle change. Some respondents reported that they had changed their job (n=4), reduced time or quit their job (n=9), or moved closer to a hospital (n=1).

Discussion

Surveys of parents of pediatric patients and of pregnant patients enrolled in clinical sequencing research projects across the US show that respondents were more likely to discuss their child's GS results with their child's clinician, and that clinicians more frequently made recommendations for a change in clinical care, if their child's GS result was positive than if it was inconclusive or negative. Overall, few respondents reported that their child's clinician(s) made recommendations for medical actions based on GS results, and furthermore, few respondents reported following up on any actions that were recommended. These findings do not support the hypothesis that GS is associated with an increase in multiple cascading medical actions, which is consistent with other studies.[9,17] Given that patient-participants were receiving GS for a clinical indication, it is possible that many of them were already receiving appropriate care based on their clinical presentation prior to GS, thus changes to care plans were not needed based on GS results.

Our findings support an understanding of care for patients with rare disease that is less focused on the diagnostic odyssey, in which parents are seeking an etiologic diagnosis, and more focused on the therapeutic odyssey, in which complex diagnostic work-ups are used to address the symptoms of the child.[18] Disentangling these conceptualizations of the patient's longitudinal journey is critical to understand the clinical and personal utility of GS in the context of rare disease diagnosis and management. Parents who had at least one child with a clinical indication for GS and were considering having additional children demonstrated strong interest in reproductive counseling, supporting the claim that informing future reproductive decisions is an important element of personal utility in both indication-based and screening contexts.[19]

Some parents self-initiated actions based on their child's GS results. Responses to open-ended questions point toward parents being more patient, more understanding, and more attentive to symptom triggers following GS testing, which is consistent with qualitative research.[20] These findings suggest that parents independently use GS results to modify the ways they interact with their child at home. The broader context of the testing process, which includes interactions with geneticists and genetic counselors, may also

provide parents with important information that can be used outside of clinical care settings. Additional research should explore how changes in parental outlook and intentions following GS affect family health and well-being.

Because of the CSER consortium research design, most respondents were medically underserved or historically underrepresented in genomics research. However, individuals who responded to survey questions generally faced fewer risk factors, as assessed by sociopolitical characteristics, for being medically underserved than non-responders. Additionally, parents of children who died before follow-up responded less frequently than those whose child was alive at the time of the survey, suggesting that care recommendations are under-reported for children who passed away during the study period. CSER participants may not have responded to the survey for one of two reasons: 1) they were asked to participate but chose not to, or 2) they were not asked to complete the survey because of project-specific sampling criteria, which may have prioritized follow-up with participants who received positive results.

While the diversity of our survey sample is a strength of this study, because the survey questions were harmonized for administration across a wide range of patients, it is possible that we did not capture all recommended changes in care, and not all survey questions were equally applicable or appropriate for administration in all projects. There are many potential reasons why parents might not have followed up on recommendations, and the degree of clinical heterogeneity limited our ability to perform more complex analyses of survey responses. Notably, clinical follow-up was largely conducted during the COVID-19 pandemic. Clinic closures or families' precautionary measures might have impacted follow-up on clinical recommendations. Additionally, we relied on parent self-report of clinicians' recommendations and did not validate against clinician-reported recommendations or documentation in patients' EMRs. However, we sampled across a broader array of clinical contexts and populations than would have been possible through EMR review. Additionally, since patients were undergoing a complex diagnostic process simultaneously with GS, we cannot parse the effects of GS results from other aspects of the diagnostic workup, including interaction with geneticists and genetic counselors, or results of other tests (although surveys asked about actions "based on the results of the genetic testing").

Overall, our findings suggest that GS results can lead to recommendations for follow up care in patients with diverse, complex medical conditions, but that they may not prompt large increases in the quantity of care received, especially for children who already have a management plan in place. However, prognostic information garnered through GS may be beneficial to patients and families even if it does not lead to additional medical actions.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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The NIH had no role in the design and conduct of the study.

Abbreviations:

GS	Genomic sequencing
RMA	recommended medical actions
PIA	parent-initiated actions

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What's Known on This Subject

Genomic sequencing has demonstrated usefulness as a diagnostic tool for infant and pediatric patients with the potential to guide clinicians' recommendations for follow-up care and provide information to patients' parents that may be personally actionable.

What This Study Adds

In diverse pediatric care settings, parental surveys suggest GS prompted moderate levels of clinical recommendations, most frequently following positive results, and potential personal benefits such as improved communication and understanding without other major changes in lifestyle or behavior.

Table 1.

Clinical Sequencing Evidence-Generating Research Consortium (CSER) sites with pediatric patients

Project	Patient Population	RMA surveys completed, n (response rate)	PIA surveys completed, n (response rate)
KidsCanSeq[a] (Baylor College of Medicine)	Pediatric cancer	136 (27.5%)	N/A
SouthSeq[b] (HudsonAlpha)	Critically ill newborns	128 (20.1%)	N/A
NYCKidSeq[c] (Mount Sinai)	Pediatric patients with undiagnosed disorders (neurologic, immunologic, and cardiac)	457 (90.1%)	457 (90.1%)
P^{3}EGS[d] (University of California, San Francisco)	Prenatal and pediatric patients with undiagnosed developmental disorders or structural anomalies	459 (55.4%)	449 (54.2%)
NCGENES 2[e] (University of North Carolina)	Pediatric patients with undiagnosed neurodevelopmental disorders	7 (50.0%)	7 (50.0%)
Total		1,187	913

N/A, not applicable because questions not asked in project surveys

[a] Surveys were matched at the provider level; when a provider had a patient with a positive result who received a survey, their next patient with a negative finding also received a survey.

[b] Surveys were administered to all participants who provided consent.

[c] Surveys were administered to all parent participants whose child was enrolled, regardless of the child's age (i.e., some children were age 18 and older).

[d] Surveys were administered to all consented participants; modified versions of the follow up survey were administered for both pediatric and prenatal patients.

[e] Survey administration was randomized at the recruitment site level, clinic level (pediatric genetics, pediatric neurology), and by served/underserved and represented/underrepresented status of eligible participants.

Table 2.

Respondent characteristics

	RMA or PIA complete	RMA & PIA incomplete	
	(n = 1,188)	(n = 1,293)	p-value
Sequencing modality			0.002
Exome	602 (50.7%)	734 (56.8%)	
Genome	586 (49.3%)	559 (43.2%)	
Respondent			<0.001
Parent of pediatric patient	1,010 (85.0%)	1,165 (90.1%)	
Pregnant patient	178 (15.0%)	128 (9.9%)	
Proband's vital status			0.001
Alive	1,038 (87.6%)	1,068 (83.0%)	
Deceased	147 (12.4%)	219 (17.0%)	
Language in which survey completed [a]			
English	1,018 (86.2%)	N/A	
Spanish	163 (13.8%)	N/A	
Child race and ethnicity [b]			0.036
American Indian, Native American, or Alaska Native	3 (0.4%)	3 (0.5%)	
Asian	31 (4.3%)	12 (2.0%)	
Black or African American	115 (15.9%)	103 (17.2%)	
Hispanic or Latino	287 (39.6%)	199 (33.3%)	
Native Hawaiian or Pacific Islander	1 (0.1%)	0 (0.0%)	
Middle Eastern or North African/Mediterranean	3 (0.4%)	4 (0.7%)	
White or European American	219 (30.2%)	208 (34.8%)	
Multiracial[c]	50 (6.9%)	46 (7.7%)	
Prefer not to answer	6 (0.8%)	14 (2.3%)	
Unknown/none fully describe	9 (1.2%)	9 (1.5%)	
CSER Underserved Framework			
Language barrier (does not speak English well (by self-report) or prefers to speak a language other than English with their healthcare provider)[d]	185 (16.9%)	171 (19.4%)	0.155
Income (household income is at or below the poverty line, as defined by the Department of Health and Human Services)[e]	403 (43.9%)	372 (54.0%)	<0.001
Insurance (does not have health insurance)[f]	5 (0.4%)	29 (3.0%)	<0.001
Residence (has a zip code that is listed in the Federal Office of Rural Health Policy list of rural zip codes)[g]	96 (8.1%)	223 (19.5%)	<0.001
Economic or geographic (meets one or more of the following underserved criteria: Insurance or Residence)[h]	461 (49.1%)	531 (66.3%)	<0.001
Barriers to access (meets one or more of the following underserved criteria: Language barrier or Economic or geographic) [i]	516 (53.1%)	591 (69.3%)	<0.001

	RMA or PIA complete	RMA & PIA incomplete	
	(n = 1,188)	**(n = 1,293)**	**p-value**
Race (self-identifies as any race other than "White or European American")[j]	505 (69.8%)	390 (65.2%)	0.079
Ethnicity (self-identifies as Hispanic/Latino(a))[k]	297 (41.0%)	209 (34.9%)	0.024
Total population at risk of being underserved or underrepresented (meets one or more of the following underserved criteria: Barriers to access, Race, or Ethnicity)[l]	716 (84.4%)	722 (88.4%)	0.019

N/A, not applicable

[a] n=7 missing;

[b] parent-reported, n=1,159 missing;

[c] includes American Indian, Native American, Alaska Native + Asian (n=1); American Indian, Native American, Alaska Native + Asian + White or European American + Hispanic/Latino(a) (n=2); American Indian, Native American, Alaska Native + Black or African American (n=4); American Indian, Native American, Alaska Native + Black or African American + White or European American (n=1); American Indian, Native American, Alaska Native + Native Hawaiian/Pacific Islander + White or European American + Hispanic/Latino(a) (n=1); American Indian, Native American, Alaska Native + White or European American (n=10); American Indian, Native American, Alaska Native + White or European American + Hispanic/Latino(a) (n=1); Asian + Black or African American + White or European American (n=1); Asian + Black or African American + White or European American + Hispanic/Latino(a) (n=1); Asian + Middle Eastern of North African/Mediterranean (n=1); Asian + White or European American (n=21); Asian + White or European American + Hispanic/Latino(a) (n=3); Black or African American + Native Hawaiian/Pacific Islander (n=1); Black or African American + White or European American (n=33); Black or African American + White or European American + Hispanic/Latino(a) (n=9); Native Hawaiian/Pacific Islander + White or European American (n=1); White or European American + Middle Eastern of North African/Mediterranean (n=5)

[d] n=503 missing;

[e] n=873 missing;

[f] n=360 missing;

[g] n=157 missing;

[h] n=741 missing;

[i] n=657 missing;

[j] n=1,159 missing;

[k] n = 1159 missing;

[l] n=816 missing

Table 3.

Parent-reported clinical communication and recommendations after receiving their child's genomic sequencing results

	Total	Positive	Inconclusive	Negative	
	(n= 1,187)	(n = 242)	(n = 407)	(n = 538)	p-value
Discussed genetic test results with child's clinician [a]					<0.001
Yes	833 (70.3%)	204 (84.3%)	256 (63.1%)	373 (69.5%)	
Not yet but I plan to	237 (20.0%)	33 (13.6%)	103 (25.4%)	101 (18.8%)	
No and I don't plan to	115 (9.7%)	5 (2.1%)	47 (11.6%)	63 (11.7%)	
If discussed results, clinician shared with					
Primary care provider/pediatrician	377 (45.3%)	109 (53.4%)	114 (44.5%)	154 (41.3%)	0.019
Neurologist	285 (34.2%)	76 (37.3%)	107 (41.8%)	102 (27.3%)	0.001
Oncologist	241 (28.9%)	61 (29.9%)	68 (26.6%)	112 (30.0%)	0.604
Cardiologist	23 (2.8%)	7 (3.4%)	11 (4.3%)	5 (1.3%)	0.067
Other specialist(s)	179 (21.5%)	62 (30.4%)	38 (14.8%)	79 (21.2%)	<0.001
If discussed, clinician made recommendations to change current care [b]					<0.001
Yes	154 (18.8%)	79 (39.1%)	31 (12.4%)	44 (11.9%)	
No	620 (75.5%)	109 (54.0%)	208 (83.2%)	303 (82.1%)	
Don't know/don't remember	47 (5.7%)	14 (6.9%)	11 (4.4%)	22 (6.0%)	

[a] n=2 missing;

[b] n=12 missing

Table 4.

Clinicians' recommendations based on child's GS result among parents who discussed results with their child's clinician and reported that a recommendation was made (n=154)

Recommendation(s)		Frequency	Percent
Medication	Start	10	6.5
	Stop	5	3.2
	Change	7	4.6
Additional non-genomic medical tests for screening, monitoring, or diagnosis	Start	25	16.2
	Stop	1	0.7
	Change	2	1.3
Referrals to consult with other doctors or specialist	Yes	19	12.3
	No	-	-
	Stop seeing specialist	-	-
Referral to a non-MD health professional	New consultation - Audiology	7	4.6
	New consultation - Dental	-	-
	New consultation - Genetic counselor	2	1.3
	New consultation - Psychologist	1	0.7
	New consultation - Other	1	0.7
Referral for mental health support	Mental health	4	2.6
	Social support	3	2.0
	Palliative care	2	1.3
Referral for therapeutic services	Speech therapy	4	2.6
	Occupational therapy	4	2.6
	Physical therapy	4	2.6
	Other	1	0.7
Lifestyle changes	Change diet	18	11.7
	Change exercise	7	4.6
	Start taking vitamins and supplements	13	8.4
	Other	3	2.0
Recommended change reported but none of the above categories checked		81	52.6