

Evaluating parental personal utility of pediatric genetic and genomic testing in a diverse, multilingual population

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Summary

There is increasing evidence of the clinical utility of genetic and genomic testing (GT); however, factors influencing personal utility of GT, especially in diverse, multilingual populations, remain unclear. We explored these factors in a diverse cohort of parents/guardians (participants) whose children received clinical GT through the NYCKidSeq program. A total of 847 participants completed surveys at baseline, post-results disclosure, and 6 months (6m) post-results. The largest population groups were Hispanic/Latino(a) (48%), White/European American (24%), and Black/African American (16%). Personal utility was assessed using the Personal Utility (PrU) scale, adapted for pediatric populations and included on the surveys. Three PrU subscales were identified using factor analysis: practical, educational, and parental psychological utility. Overall personal utility summary score and the three subscales significantly decreased after receiving results and over time. Hispanic/Latino(a) participants identified greater overall personal utility than European American and African American participants at all time points ($p < 0.001$) as did participants whose children received positive/likely positive results compared with those with negative and uncertain results (post-results: $p < 0.001$ and $p < 0.001$; 6m post-results: $p = 0.002$ and $p < 0.001$, respectively). Post-results, higher subscale scores were associated with lower education levels (practical, parental psychological: $p \leq 0.02$) and higher levels of trust in the healthcare system (practical, parental psychological: $p \leq 0.04$). These findings help to understand the perspectives of diverse parents/guardians, which is critical to tailoring pre- and post-test counseling across a variety of populations and clinical settings.

Introduction

Genetic and genomic testing (GT) has revolutionized our understanding of the genetic basis of disease and has emerged as a valuable tool in pediatric clinical settings. Advances in sequencing technology have increased access to genetic testing within a clinically useful time frame, providing families and healthcare providers with the opportunity for a molecular diagnosis, tailored treatment, screening recommendations, and cascade testing.¹ There is increasing evidence of the utility of GT in clinical care, or clinical utility, with previous studies demonstrating how GT helps provide a diagnosis, identify dis-

ease risk, and guide medical interventions.^{2,3} However, the value of GT beyond, or independent from, clinical utility, is not as well understood. Personal utility is a concept that takes into account the non-medical value of GT information to the patient or the patients' family members.⁴ Reported elements of personal utility associated with genetic testing encompass personal knowledge, understanding of a health condition (i.e., diagnosis, prognosis, recurrence risk), altruism, and coping ability.⁴⁻⁹

Unlike clinical utility, there is still limited information on the factors that contribute to the personal utility of GT. The few studies that have explored patients' and

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<https://doi.org/10.1016/j.xhgg.2024.100321>.

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parents' perspectives on the impact of GT, suggest that patients and parents experience utilities and disutilities across clinical, psychological, and pragmatic domains. For example, for both patients' and parents', utilities may include benefits of early disease detection, satisfaction of curiosity, and future planning. On the other hand, disutilities can include blame from family members for passing on a health condition and discrimination from employers, schools, or insurance.^{10–13} In the context of a diagnostic (positive) genetic result, personal utility has been linked to ending the diagnostic odyssey, reducing emotional and financial burdens, and yielding information relevant to making reproductive decisions and accessing appropriate services and support.^{4,7,8,14–16} For non-diagnostic (uncertain and/or negative) genetic results, personal utility can be related to ruling out certain conditions, giving parents confidence that they have exhausted diagnosis testing for their child, and allowing families to pause the diagnostic odyssey to focus on symptom management.^{7,17} Although there is overlap, it is important to note the differences in motivations and utilities between adult patients and parents whose children undergo GT. One qualitative study of parents whose child received genetic testing reported that many parents worried about the possibility of a lethal diagnosis, suggesting heightened anxiety around GT for parents when compared with adult patients.¹⁷ Overall, however, the extent to which previously published findings on personal utility of GT apply to a diversity of patients and families remains unclear, since the majority of studies primarily involved highly educated, non-Hispanic, White parents.^{7,8,17–20}

Sociodemographic factors, including education level, income, primary language, and healthcare system factors related to racial bias and access to insurance, all have the potential to impact the personal utility of GT.^{6,21–23} It is therefore critical to evaluate parents' personal utility of GT in a diverse pediatric patient population to gain a more thorough and nuanced understanding of the value parents place on GT for their child. The NYCKidSeq research program enrolled diverse and medically underserved children with suspected genetic conditions who underwent GT and evaluated the clinical and personal utility of the genetic testing.^{24,25} NYCKidSeq was jointly funded by the National Human Genome Research Institute and the National Institute on Minority Health and Health Disparities and was one of seven national clinical sites that were part of the Clinical Sequencing Evidence-Generating Research consortium.²⁶ We previously reported on the diagnostic yield of GT³ as well as the impact of a digital application on parents' understanding of their child's GT results.²⁷ In the present study, we describe participants' assessment of the personal utility of the testing at baseline, post-results disclosure, and 6-months (6m) post-results disclosure, which highlights the factors that contribute to personal utility in a diverse pediatric patient population.

Materials and methods

Overview

The NYCKidSeq program comprised two research studies, the NYCKidSeq randomized control trial (NCT03738098), which evaluated the use of a digital platform designed to facilitate results disclosure, and the TeleKidSeq pilot study, which assessed the benefits screensharing during results disclosure. These studies have been described previously by Odgis et al., in 2021, and Sebastian et al., in 2023, respectively.^{24,25} Recruitment for the NYCKidSeq and TeleKidSeq studies occurred at two New York City health systems, Mount Sinai Medical Center and Albert Einstein/Montefiore Medical Center. This study includes children and their parents/legal guardians from both study cohorts. The institutional review boards at the Icahn School of Medicine at Mount Sinai and the Albert Einstein College of Medicine approved the studies, and all participants provided written informed consent.

Study population

Study participants are parents and legal guardians of the children (<21 years) who received GT through the NYCKidSeq program. These children had a suspected genetic etiology for their neurologic, cardiac, or immunologic condition, had not had genetic testing or had a previously uninformative genetic test, and had not undergone genetic counseling in the 3 months preceding enrollment. They underwent clinical GT, which included genome sequencing (GS) and targeted gene panels (TGPs) for a subset ($n = 642$). For those who did receive a TGP, one or multiple panel(s) were run based on the child's phenotype. GT was performed by Clinical Laboratory Improvement Amendments (CLIA)-certified and New York State approved genetic testing laboratories.³ Study genetic counselors (GCs) provided parents/legal guardians (participants) with pre-test genetic counseling to discuss the risks, benefits, and limitations of GT and post-test counseling to review their child's results and care recommendations approved by their referring provider. Participants were required to be fluent in English and/or Spanish to participate in the study. Pre- and post-test genetic counseling was conducted with the use of a Spanish interpreter per participant preference. The NYCKidSeq study developed a digital platform designed to facilitate the communication of GT results, called GUÍA (Genomic Understanding, Information, and Awareness application). This platform was designed to facilitate results disclosure of GT results in English and Spanish with the use of text and images.²⁸ Participants enrolled in NYCKidSeq were randomized to post-test counseling with or without the use of GUÍA. Participants either received the results of the GT in person ($n = 155$, 16.4%) or via telehealth ($n = 789$, 83.6%). Participants who received the results via telehealth either received their results with ($n = 192$, 20.3%) or without ($n = 597$, 63.2%) screen sharing capabilities. The results of GT were entered into the child's electronic medical record. Participants completed surveys before genetic testing (baseline), post-results disclosure (typically immediately after results disclosure, with a maximum of 1 month post-results disclosure), and 6m post-results disclosure. Bilingual research coordinators administered surveys in English or Spanish per participant preference, and recorded responses in REDCap (Research Electronic Data Capture), a secure, web-based software platform.^{29,30} Participants received an incentive (up to \$80) for completion of all components of the study.

Personal utility measure

We assessed participants' personal utility of GT across the three time points using the 19-item Personal Utility (PrU) scale⁹ adapted for parents/legal guardians of pediatric populations (see Table S1 for PrU questions).³¹ The measure evaluated expected personal utility at baseline, and then actual personal utility post-results disclosure. Participants who indicated a preference to complete the surveys in Spanish received a Spanish translation of the same PrU scale. Participants responded to each item using a seven-point scale ranging from "not useful at all" (1) to "extremely useful" (7) (see Table S2).

Analytic sample

Participants enrolled in the NYCKidSeq program were eligible for this study unless they were enrolled in the lead-in phase ($n = 37$), did not complete all three surveys ($n = 44$), completed the post-result survey >1 month from results disclosure ($n = 1$), or their child's case-level clinical interpretation was amended between the two post-results disclosure surveys ($n = 1$). After exclusion of participants who were lost to follow-up ($n = 79$), the final analytic sample consisted of 944 individuals' post-results disclosure and 847 6m post-results disclosure.

Sociodemographic and clinical variables

We collected participants' sociodemographic data at baseline, which included self-reported race and ethnicity, education level, and health insurance type. In addition, we captured the following clinical information on enrolled children: primary phenotype (neurologic, cardiac, immunologic), sex assigned at birth, and case-level interpretation of GT. At baseline, participants completed the validated four-item Brief Health Literacy Screening Tool³² and the validated nine-item Health Care Distrust scale, comprising two subscales (competence and values).³³ See Table S3 for all population characteristics that were collected.

Participants were asked to select one or more of nine population descriptors that applied to them, and these were transformed into population groups used for analysis. Due to the wording of the question, the survey instrument failed to differentiate between legal guardians and biological parents when asking for population descriptors. Therefore, data were collected for biological parents only ($n = 904$), legal guardians were not assigned population groups ($n = 40$). Participants who selected Hispanic/Latino(a) (H/L) were assigned as H/L regardless of any other population designation made and collapsed into one category ($n = 435$). Participants who selected more than one population descriptor, not including H/L, were collapsed and assigned as "More than one population" ($n = 17$). Participants who selected just one population descriptor, not including H/L, were assigned as those population groups; American Indian ($n = 3$), Asian ($n = 55$), Black or African American (AA) ($n = 141$), Middle Eastern or North African/Mediterranean ($n = 10$), and White or European American (EA) ($n = 217$). Participants also had the option to select a population descriptor of other ($n = 9$), prefer not to answer ($n = 11$), or unknown/none of these fully describe the child's mother/father ($n = 6$).

Education level was collapsed into four categories: less than a high school degree (HS) ($n = 188$), HS/General Education Development (GED)/post-HS training ($n = 422$) college graduate ($n = 190$), and greater than a college degree ($N = 161$). Case-level clinical interpretation of genetic results was determined by GCs as positive, likely positive, uncertain, or negative following criteria described previously.³ Positive and likely positive categories were

combined in the present study due to similarities in results disclosure. Mode of result delivery was collapsed into either in-person ($n = 197$) or telehealth, which consisted of those who were met over teleconference ($n = 740$) and over the phone ($n = 7$).

We assessed participants' understanding of their child's GT results using two novel measures developed for the NYCKidSeq study.²⁷ Perceived understanding of the results was measured on a Likert scale of 1 ("very little or none of it") to 5 ("almost all or all of it"). For our analyses, survey response 1 was eliminated due to a low number of responses ($N = 11$) and the survey responses of 2 ($N = 13$) and 3 ($N = 81$) were collapsed into a single category designated as level 1. Survey response 4 ($N = 261$) was categorized as level 2. Finally, survey response 5 ($N = 578$) was categorized as level 3. Objective understanding was assessed through four questions (Table S2) asked to participants and GCs following the return of results. An objective understanding summary score was calculated by totaling the agreement of participant responses with the GCs. The objective understanding summary score ranged from 0 as the lowest possible score to 4 as the highest.

Statistical analyses

A previously published factor analysis of the adult version of the PrU identified three subscales: self-knowledge (6 items), reproductive planning (2 items), and practical benefits (6 items).³¹ We conducted a factor analysis for the adapted pediatric PrU, removing two items ("Use for testing a future pregnancy, if appropriate" and "Inform my child's decisions about having children") *a priori* since these items were not applicable to the entire study cohort (e.g., the question about future pregnancy was not relevant to legal guardians). The remaining 15 items loaded on to 3 subscales, which we described as: "practical utility" (8 items, possible subscale score range: 8–56), "educational utility" (5 items, possible subscale score range: 5–35), and "parental psychological utility" (2 items, possible subscale score range: 2–14; Table S1) based on the utilities that comprised each category. Internal consistency was high among the analytical study sample for each subscale ("practical utility" Cronbach's $\alpha = 0.94$, "educational utility" Cronbach's $\alpha = 0.87$, and "parental psychological" Cronbach's $\alpha = 0.97$). For each time point, we calculated individual subscale scores and constructed an overall personal utility score by adding responses for all 15 items. Participants with missing responses were not included in the subscale and overall score calculations.

We employed descriptive statistics to characterize all study variables within the analytic sample. One-way analysis of variance (ANOVA) was used to evaluate the mean of PrU subscale scores and summary score separately within the three time points. Bivariate analysis was used to assess the association of sociodemographic and clinical characteristics with the PrU summary score at post-results disclosure and 6m post-results disclosure time points (see Table S4). Of these, we identified sociodemographic and clinical factors to use as covariates in the analyses, including: survey language, education level, insurance type, case-level clinical interpretation of genetic results, history of previous genetic testing, health literacy score, and healthcare distrust scores (values subscale and competence subscale). Population group was not included as a covariate in our models due to several categories having small sample sizes. Baseline surveys were completed pre-test, therefore case-level clinical interpretation was not known and not included in baseline models.

Using multivariate linear regression, we evaluated the association of the covariates with the three PrU subscale scores and the

Table 1. Participant characteristics for NYCKidSeq, TeleKidSeq, and the total sample

Participant characteristic, N (%)	NYCKidSeq (n = 551)	TeleKidSeq (n = 393)	Total sample (n = 944)
Age of parent at baseline (mean, SD, range) ^a	41.1 (9.0) 20.9–81.5	40.5 (9.2) 21.3–71.7	40.89 (9.1) 20.9–81.5
Relationship to child			
Mother	488 (88.6)	335 (85.2)	823 (87.2)
Father	43 (7.8)	38 (9.7)	81 (8.6)
Legal guardian	20 (3.6)	20 (5.1)	40 (4.2)
Recruitment site			
Montefiore Medical Center	187 (34.0)	134 (34.1)	321 (34.0)
Mount Sinai Health System	364 (66.0)	259 (66.0)	623 (66.0)
Previous genetic testing ^b	222 (40.3)	146 (37.2)	368 (39.0)
Self-reported race and ethnicity ^c			
American Indian	2 (0.4)	1 (0.3)	3 (0.3)
Asian	38 (7.2)	17 (4.6)	55 (6.1)
Black or African American	84 (15.8)	57 (15.3)	141 (15.6)
Hispanic/Latino(a)	245 (46.1)	190 (51.0)	435 (48.1)
Middle Eastern or North African/Mediterranean	8 (1.5)	2 (0.5)	10 (1.1)
White or European American	129 (24.3)	88 (23.6)	217 (24.0)
More than one population	9 (1.7)	8 (2.1)	17 (1.9)
Other	4 (0.8)	5 (1.3)	9 (1.0)
Prefer not to answer	6 (1.1)	5 (1.3)	11 (1.2)
Unknown/None of these fully describe my child	6 (1.1)	0 (0.00)	6 (0.7)
Survey conducted in Spanish	121 (22.0)	90 (23.0)	211 (22.4)
Interpreter present at post-test	107 (19.4)	85 (21.6)	192 (20.3)
How was the survey administered? ^d			
Phone	128 (23.2)	157 (40.1)	285 (30.2)
In-person	148 (26.9)	5 (1.3)	153 (16.2)
Videoconference	275 (49.9)	230 (58.7)	505 (53.6)
Education level ^e			
< HS	100 (18.2)	68 (17.4)	168 (17.9)
HS/GED/post-HS training	242 (44.1)	180 (45.9)	422 (44.9)
College graduate	114 (20.8)	76 (19.4)	190 (20.2)
> College graduate	93 (17.0)	68 (17.4)	161 (17.1)
Public insurance (Medicaid/Medicare)	357 (64.8)	249 (63.4)	606 (64.2)
MUAP (residence in an HRSA defined “medically underserved area”)	297 (53.9)	189 (48.1)	486 (51.5)
200% below NYC federal poverty level ^f	230 (41.7)	181 (46.1)	411 (43.5)
Brief Health Literacy Score (mean, SD, range)	16.8 (3.6) 4.0–20.0	17.1 (3.6) 4.0–20.0	17.0 (3.6) 4.0–20.0
Healthcare Distrust Values Subscale Score (mean, SD, range) ^d	15.6 (3.9) 5.0–25.0	15.8 (3.7) 5.0–25.0	15.7 (3.8) 5.0–25.0
Healthcare Distrust Competence Subscale Score (mean, SD, range) ^d	10.91 (2.9) 4.0–20.0	10.97 (3.0) 4.0–20.0	10.94 (2.9) 4.0–20.0
Child characteristic, N (%)			

(Continued on next page)

Table 1. Continued

Participant characteristic, N (%)	NYCKidSeq (n = 551)	TeleKidSeq (n = 393)	Total sample (n = 944)
Age of child at baseline in years (mean, SD, range)	9.8 (5.8) 0.1–22.0	9.2 (5.7) 0.1–22.0	9.6 (5.7) 0.1–22.0
Child sex assigned at birth			
Female	202 (36.7)	151 (38.4)	353 (37.4)
Male	349 (63.3)	242 (61.6)	591 (62.6)
Primary phenotype			
Cardiac	21 (3.8)	20 (5.1)	41 (4.3)
Immunologic	29 (5.3)	19 (4.8)	48 (5.1)
Neurologic	501 (90.9)	354 (90.1)	855 (90.6)
Case-level clinical interpretation of genetic test result			
Positive/likely positive	91 (16.5)	73 (18.6)	164 (17.3)
Uncertain	317 (57.5)	165 (42.0)	482 (51.1)
Negative	143 (26.0)	155 (39.4)	298 (31.6)

SD, standard deviation; HS, high school; GED, General Education Development Test; NYC, New York City; HRSA, Health Resources & Services Administration.

^aDue to missing data, NYCKidSeq *n* = 549, TeleKidSeq *n* = 391, total sample *n* = 940.

^bEleven participants answered, “Don’t Know”: NYCKidSeq, *n* = 6, TeleKidSeq, *n* = 5.

^cRace and ethnicity was not collected for legal guardians. Therefore, NYCKidSeq *n* = 531, TeleKidSeq *n* = 373, total sample *n* = 904.

^dDue to missing data, NYCKidSeq *n* = 551, TeleKidSeq *n* = 392, total sample *n* = 943.

^eDue to missing data, NYCKidSeq *n* = 549, TeleKidSeq *n* = 392, total sample *n* = 941.

^fMissing data due to incomplete information regarding household size and approximate household income.

PrU summary score. Separate analyses were run for each time point: baseline, post-results disclosure, and 6m post-results disclosure. To further analyze the association between population group and personal utility, we conducted ANOVAs and post-hoc Tukey tests using the overall PrU summary score across all three time points and the three largest population groups (EA, AA, and H/L). Stata version 17 was used for all analyses (2021, StataCorp LP).

To further investigate the factors that may impact personal utility among different population groups, an additional multivariate linear regression was conducted, including stratification by the three largest population groups. Models were conducted at two time points, post-results disclosure, and 6m post-results disclosure, and included mode of result delivery (in person vs. telehealth), perceived understanding, objective understanding, and genetic counselor as additional covariates. Stata version 17 was used for all analyses (2021, StataCorp LP).

Results

Participants and children characteristics

The mean age of study participants was 40.9 years and the majority were mothers (87.2%) (Table 1). The three largest population groups were H/L (48.1%), EA (24.0%), and AA (15.6%), followed by Asian (6.1%), Middle Eastern or North African/Mediterranean (1.1%), American Indian (0.3%), more than one population (1.9%), prefer not to answer (1.2%), other (1.0%), and unknown/none of these (0.7%). Baseline surveys were conducted in Spanish for 22.4% of participants. Over half had less than a college degree (62.7%), 43.5% had a household income reported that was at or below 200% of the New York City (NYC) Federal

poverty level, and 51.5% resided within a Health Resources and Services Administration (HRSA)-defined medically underserved area (<https://data.hrsa.gov>).³⁴ The mean age of enrolled children was 9.6 years (range 1 month–21 years) and 62.6% were male. The majority had a primary neurological phenotype (90.6%). Of the children, 16.5% received positive/likely positive results, 57.3% received uncertain results, and 26.0% received negative results. Population characteristics of participants enrolled in the NYCKidSeq and TeleKidSeq studies are also displayed in Table 1.

Three domains of personal utility of pediatric genomic testing

We assessed participants’ overall perceptions of the personal utility of the GT at baseline, post-results disclosure, and 6m post-results disclosure (with an average completion time of 6.4 months) using the summary score of the PrU measure. The possible range for the summary score was 15–105, with higher scores indicating greater personal utility. Overall, PrU summary scores were high across the three time points. However, PrU scores significantly decreased over time, from an average summary score of 86.2 (standard deviation [SD] = 15.1) at baseline to 81.6 (SD = 20.7) post-results disclosure to 76.4 (SD = 22.7) 6m post-results disclosure (*p* < 0.001; Figure 2A).

We conducted a factor analysis of the PrU measure, identifying three subscales, or domains, of personal utility, which we describe as practical utility, educational utility, and parental psychological utility (Figure 1). Practical utility encompasses how GT results can be helpful in

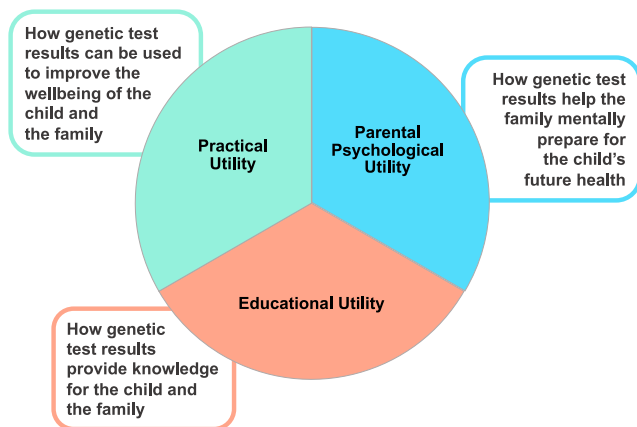


Figure 1. Domains of personal utility of genetic test results
The results of the factor analysis of the Personal Utility (PrU) scale adapted for parents/legal guardians of pediatric populations identified three subscales: practical utility, educational utility, and parental psychological utility.

supporting the child's well-being and development. Educational utility captures GT's usefulness in providing families with sought-after information and knowledge about their child's condition. Lastly, parental psychological utility involves how the GT may help families mentally prepare for the future, mainly in relation to their child's health. For practical utility, the mean subscale score was 44.5 (SD = 9.4; possible range 8–56) at baseline, 41.2 (SD = 13.1) at post-results disclosure, and 38.1 (SD = 14.0) at 6m post-results disclosure. The educational utility mean subscale score was 30.0 (SD = 4.6; possible range 5–35) at baseline, 29.5 (SD = 5.7) at post-results disclosure, and 28.1 (SD = 6.8) at 6m post-results disclosure. The mean parental psychological utility subscale score was 11.7 (SD = 2.3; possible subscale range 2–14) at baseline, 10.9 (SD = 3.3) at post-results disclosure, and 10.2 (SD = 3.6) at 6m post-results disclosure. We also evaluated the impact of time on the individual subscales and observed that, like the summary score, the average subscale scores significantly decreased over time (practical utility $p \leq 0.001$, educational utility $p \leq 0.001$, parental psychological utility $p \leq 0.001$; [Figures 2B–2D](#)).

Socioeconomic and clinical factors associated with personal utility of pediatric genomic testing

To explore how socioeconomic and clinical factors impact participants' views on the personal utility of GT, we performed multivariate linear regression models across the three study time points. This analysis included the PrU summary score as well as the three PrU subscales, evaluating six participant characteristics: survey language, education level, insurance type, health literacy, healthcare distrust (values and competence), case-level clinical interpretation of GT results, and previous genetic testing. We also conducted a comparison of PrU summary scores among the three largest AA, H/L, and EA population

groups in our study. We excluded other populations from this analysis due to their limited sample size.

Personal utility of pediatric genomic testing: Summary score

We observed that the PrU summary score was associated with participants' education level, health literacy score, healthcare distrust, and the case-level interpretation of the GT results ([Table 2](#)). At baseline, participants with HS/GED/post-HS training expected significantly greater personal utility than those with less than an HS education ($b = 3.57, p = 0.016$); however, there were no differences between those with less than HS education and those with a college degree or greater. Once participants received their results, the association between education level and personal utility abated. At post-results disclosure, higher health literacy levels were associated with lower personal utility summary scores ($b = -0.47, p = 0.024$). We also observed that higher levels of distrust in the competence of the healthcare system were associated with lower personal utility at baseline and post-results disclosure ($b = -0.67, p = 0.003$ and $b = -0.65, p = 0.030$, respectively), with similarly trending results at 6m post-results disclosure ($b = -0.68, p = 0.052$). Furthermore, the classification of the clinical results significantly impacted participants' views on the personal utility of the GT. Those receiving a positive/likely positive result had higher PrU summary scores post-results disclosure compared with those receiving a negative result ($b = 13.36, p < 0.001$) and to those receiving an uncertain result ($b = 14.04, p < 0.001$). This trend persisted 6m post-results disclosure ($b = 7.11, p = 0.002$ and $b = 10.56, p < 0.001$, respectively). Conversely, at 6m post-results disclosure, those with uncertain results had lower personal utility summary scores when compared with those receiving a negative result ($b = -3.85, p = 0.025$). Insurance type, previous genetic testing, and the healthcare distrust values subscale were not associated with the PrU summary score.

To gain a deeper understanding of how different populations perceive the personal utility of GT, we compared PrU summary scores between populations and identified significant differences between the groups using ANOVA and post-hoc Tukey tests (baseline: $p < 0.001$; post-results disclosure: $p < 0.001$, 6m post-results disclosure: $p < 0.001$; [Figure S2](#)). Overall, H/L participants identified greater personal utility of the GT than EA and AA participants, a consistent finding across the three study time points ([Figure S2](#)). The PrU summary scores were not significantly different between AA and EA groups at either post-results disclosure time points.

To further explore what factors affecting personal utility might be driving differences among the three largest population groups in our study, we conducted a multivariate linear regression. We stratified by population groups and included a number of covariates ([Tables S5 and S6](#)). At post-results disclosure, perceived understanding, objective understanding, insurance type, healthcare distrust, and

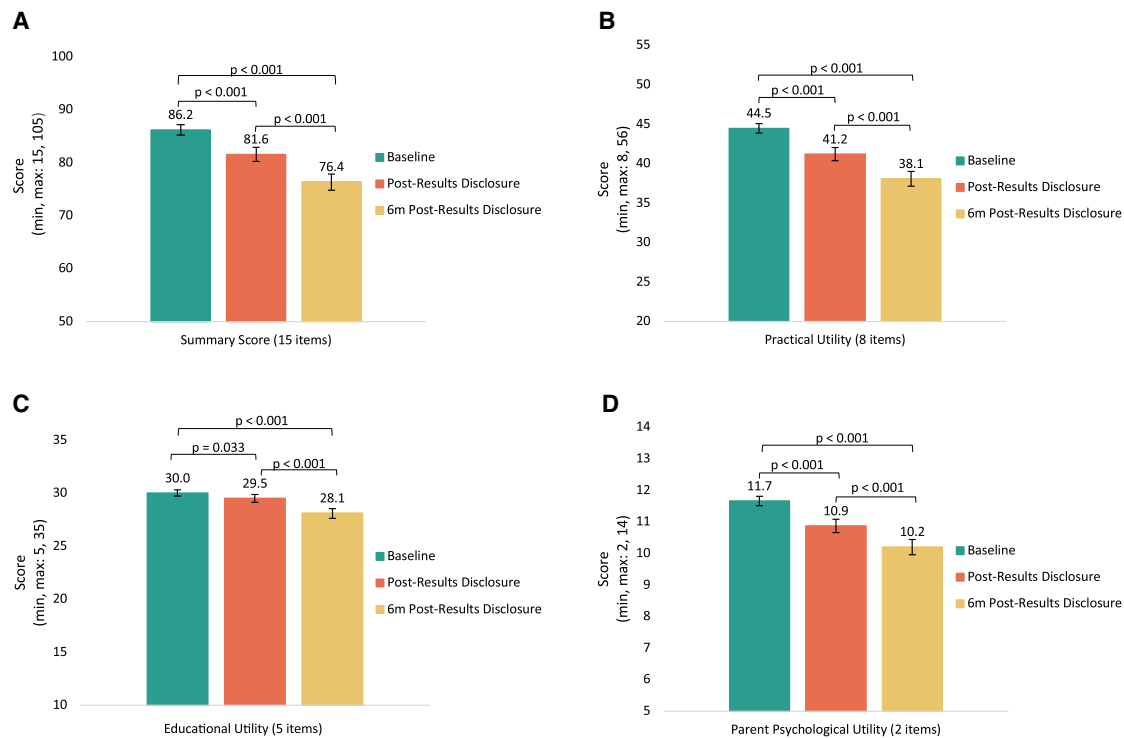


Figure 2. Comparison of personal utility of genomic results assessed at baseline, post-results disclosure, and 6m post-results disclosure

Error bars represent 95% confidence intervals. One-way ANOVAs analyzed significant differences in PrU mean scores at baseline, post-results disclosure, and 6m post-results disclosure using (A) PrU summary scores, (B) the practical utility subscale, (C) the educational utility subscale, and (D) the parental psychological utility subscale. *p* values were calculated from post-hoc Tukey HSD analyses.

case-level clinical interpretation were all found to have associations with overall personal utility at the population level. In the full sample, higher perceived understanding was associated with greater personal utility summary scores (level 2 vs. level 1: $b = 6.8, p = 0.004$; level 3 vs. level 1: $b = 10.2, p < 0.001$), which persisted across the three population groups. Conversely, higher objective understanding was associated with lower personal utility summary scores ($b = -2.3, p < 0.001$). This association was also found for AA ($b = -5.0, p = 0.008$) and H/L ($b = -1.7, p = 0.038$) participants, but not for EA participants. Participants who received positive/likely positive results reported higher personal utility than those who received negative results across the three population groups (EA: $b = 15.09, p < 0.001$; AA: $b = 20.479, p = 0.001$; H/L: $b = 13.95, p < 0.001$). Only EA participants demonstrated a negative association between personal utility and distrust in the competence of the healthcare system ($b = -1.53, p = 0.014$). H/L parents with private insurance reported lower personal utility than those with public ($b = -4.95, p = 0.045$), as did those with previous genetic testing ($b = -4.26, p = 0.026$). There was no association with personal utility and mode of results disclosure in the full sample at either time point. In addition, within the population groups, there was no association between personal utility and mode of results disclosure, education level, survey language, or health literacy score. At the 6m post-results

disclosure time point, although some significant associations seen post-results disclosure persisted, most of the associations were no longer observed.

Personal utility of pediatric genomic testing: Practical utility subscale

Several socioeconomic and clinical factors were associated with the practical utility subscale, including participants' education level, healthcare distrust, insurance type, health literacy level, history of previous genetic testing, and case-level interpretation of the results (Table 3). We found that participants' level of education impacted their views on the practical utility of the testing, which varied depending on the study time point. Those with HS/GED/post-HS training reported higher expected practical utility at baseline compared with those with less than HS education ($b = 1.83, p = 0.042$), but this difference did not persist after results were disclosed. We also observed that those with higher education levels, including college graduates and those with higher than a college degree, had significantly lower practical utility scores post results disclosure compared with those with less than HS education ($b = -3.24, p = 0.027$ and $b = -4.09, p < 0.001$, respectively). At the 6m post-results time point, those with an education level higher than a college degree continued to report significantly lower practical utility scores ($b = -3.62, p = 0.046$). Across the three study time points, higher levels

Table 2. Multivariate linear regression of the personal utility summary score assessed at baseline, post-results disclosure, and 6m post-results disclosure

	Baseline (<i>n</i> = 900)		Post-results disclosure (<i>n</i> = 923)		6m Post-results disclosure (<i>n</i> = 828)	
	β (95% CI)	<i>p</i> value	β (95% CI)	<i>p</i> value	β (95% CI)	<i>p</i> value
Socioeconomic factors						
Education level						
HS/GED/post-HS training vs. < HS	3.57 (0.67, 6.47)	0.016	2.72 (−1.05, 6.50)	0.157	3.17 (−1.20, 7.53)	0.155
College graduate vs. < HS	0.35 (−3.15, 3.86)	0.843	−3.83 (−8.41, 0.75)	0.101	−0.22 (−5.56, 5.12)	0.936
> College+ vs. < HS	−1.67 (−5.48, 2.15)	0.392	−4.71 (−9.70, 0.29)	0.065	−4.19 (−10.02, 1.64)	0.159
Survey language						
Spanish vs. English	0.51 (−2.30, 3.32)	0.722	1.28 (−2.32, 4.88)	0.486	2.65 (−1.48, 6.79)	0.208
Insurance type						
Private vs. public	−1.85 (−4.17, 0.48)	0.119	−2.41 (−5.45, 0.64)	0.121	−2.51 (−6.08, 1.06)	0.168
Healthcare distrust						
Values Subscale	0.09 (−0.26, 0.43)	0.631	0.21 (−0.25, 0.67)	0.370	−0.17 (−0.70, 0.36)	0.529
Competence Subscale	−0.67 (−1.12, −0.23)	0.003	−0.65 (−1.24, −0.06)	0.030	−0.68 (−1.37, 0.01)	0.052
Health Literacy Score	−0.17 (−0.48, 0.14)	0.288	−0.47 (−0.87, −0.06)	0.024	−0.22 (−0.70, 0.25)	0.354
Clinical factors						
Case-level clinical interpretation						
Positive/likely positive vs. negative	–		13.36 (9.55, 17.18)	<0.001	7.11 (2.69, 11.52)	0.002
Uncertain vs. negative	–		−0.68 (−3.54, 2.18)	0.642	−3.85 (−7.22, −0.49)	0.025
Positive/likely positive vs. uncertain ^a	–		14.04 (10.49, 17.58)	<0.001	10.56 (6.86, 15.06)	<0.001
Previous genetic testing						
Yes vs. No	−1.46 (−3.46, 0.55)	0.154	−1.83 (−4.46, 0.80)	0.173	−2.70 (−5.78, 0.37)	0.085

HS, high school; GED, General Education Development Test.

Bold entries signify significant results.

^aPost-hoc analyses were conducted using uncertain result as the reference variable for case-level clinical interpretation.

of distrust in the competence of the healthcare system were negatively associated with practical utility (baseline: $b = -0.49$, $p = 0.001$; post results disclosure: $b = -0.46$, $p = 0.014$; 6m post-results disclosure $b = 0.43$, $p = 0.044$). In addition, those with private insurance had lower practical utility scores at baseline ($b = -1.47$, $p = 0.042$) and 6m post-results disclosure ($b = -2.30$, $p = 0.039$) compared with those with public insurance. Lastly, health literacy levels were negatively associated with practical utility at the post-results disclosure time point ($b = -0.28$, $p = 0.028$). In the context of the clinical factors, prior genetic testing was associated with lower practical utility scores at both post-results disclosure ($b = -1.69$, $p = 0.044$) and 6m post-results disclosure ($b = -2.11$, $p = 0.027$) time points. In addition, participants whose child received a positive/likely positive test result identified significantly higher practical utility of the testing compared with a negative result and with an uncertain result at post-results disclosure ($b = 8.33$, $p < 0.001$ and $b = 8.67$, $p < 0.001$, respectively) and 6m post-results

disclosure ($b = 5.27$, $p < 0.001$ and $b = 7.62$, $p < 0.001$, respectively). Uncertain results were associated with lower practical utility scores compared with negative results only at 6m post-results disclosure ($b = -2.35$, $p = 0.025$). Survey language and the healthcare distrust values subscale were not associated with practical utility.

Personal utility of pediatric genomic testing: Educational utility subscale

Educational utility subscale scores were associated with participants' education level, healthcare distrust, and the case-level interpretation of the results (Table 4). Across the three study time points, having an HS/GED/post-HS training was associated with higher educational utility scores compared with those with less than HS education (baseline: $b = 1.08$, $p = 0.018$; post-results disclosure: $b = 1.18$, $p = 0.032$; 6m post-results disclosure: $b = 1.60$, $p = 0.020$). A higher level of distrust in the competence of the healthcare system was negatively associated with educational utility at baseline ($b = -0.15$, $p = 0.029$).

Table 3. Multivariate linear regression of the practical utility subscale assessed at baseline, post-results disclosure, and 6m post-results disclosure

	Baseline (n = 913)		Post-results disclosure (n = 927)		6m Post-results disclosure (n = 831)	
	β (95% CI)	p value	β (95% CI)	p value	β (95% CI)	p value
Socioeconomic factors						
Education level						
HS/GED/post-HS training vs. <HS	1.83 (0.06, 3.61)	0.042	1.29 (-1.08, 3.65)	0.286	0.95 (-1.71, 3.61)	0.485
College graduate vs. <HS	-0.42 (-2.56, 1.72)	0.702	-3.24 (-6.10, -0.38)	0.027	-1.20 (-4.46, 2.05)	0.469
College+ vs. <HS	-1.73 (-4.07, 0.60)	0.145	-4.09 (-7.22, -0.97)	<0.001	-3.62 (-7.17, -0.06)	0.046
Survey language						
Spanish vs. English	-0.03 (-1.74, 1.67)	0.971	0.78 (-1.47, 3.03)	0.497	0.95 (-1.57, 3.47)	0.458
Insurance type						
Private vs. Public	-1.47 (-2.88, -0.05)	0.042	-1.66 (-3.56, 0.24)	0.086	-2.30 (-4.48, -0.12)	0.039
Healthcare distrust						
Values subscale	0.07 (-0.15, 0.28)	0.538	0.13 (-0.16, 0.41)	0.382	-0.14 (-0.46, 0.18)	0.397
Competence subscale	-0.49 (-0.76, -0.21)	0.001	-0.46 (-0.83, -0.10)	0.014	-0.43 (-0.84, -0.01)	0.044
Health Literacy Score	-0.14 (-0.33, 0.05)	0.161	-0.28 (-0.54, -0.03)	0.028	-0.20 (-0.49, 0.09)	0.176
Clinical factors						
Case-level clinical interpretation						
Positive/likely positive vs. negative	-		8.33 (5.95, 10.71)	<0.001	5.27 (2.57, 7.96)	<0.001
Uncertain vs. negative	-		-0.34 (-2.13, 1.45)	0.708	-2.35 (-4.40, -0.30)	0.025
Positive/likely positive vs. uncertain ^a	-		8.67 (6.95, 10.89)	<0.001	7.62 (5.12, 10.12)	<0.001
Previous genetic testing						
Yes vs. no	-1.21 (-2.44, 0.01)	0.052	-1.69 (-3.34, -0.04)	0.044	-2.11 (-3.98, -0.24)	0.027

HS, high school; GED, General Education Development Test.

Bold entries signify significant results.

^aPost-hoc analyses were conducted using uncertain result as the reference variable for case-level clinical interpretation.

Positive/likely positive results were more likely to have higher educational utility scores at post-result disclosure compared with negative and uncertain results ($b = 3.02$, $p < 0.001$ and $b = 3.61$, $p < 0.001$, respectively). Uncertain results were more likely to have lower educational utility scores than negative results at 6m post-results disclosure ($b = -1.19$, $p = 0.024$). Survey language, insurance type, health literacy score, healthcare distrust values subscale, and previous genetic testing were not associated with educational utility.

Personal utility of pediatric genomic testing: Parental psychological utility subscale

A number of socioeconomic and clinical factors were associated with parental psychological utility scores, including education level, health literacy score, healthcare distrust, survey language, and case-level interpretation of the results (Table 5). At baseline, those with HS/GED/post-HS training had higher expected parental psychological utility scores than those with less than HS ed-

ucation ($b = 0.56$, $p = 0.014$). However, this trend did not persist post-results. In fact, after results disclosure, college graduates and those with more than a college education were more likely to report lower parental psychological utility of the GT ($b = -0.74$, $p = 0.048$ and $b = -0.93$, $p = 0.024$, respectively). In addition, we observed that those with higher health literacy levels and greater distrust in the competence of the healthcare system had lower parental psychological utility scores post-results disclosure ($b = -0.06$, $p = 0.033$ and $b = -0.10$, $p = 0.037$, respectively). Parental psychological utility is the only subscale impacted by the language the survey was completed in (English or Spanish). We found that, after results disclosure, those who completed the survey in Spanish had higher parental psychological utility compared with those completing the English version of the survey ($b = 2.00$, $p < 0.001$). As with the summary score and other subscales, positive/likely positive results were positively associated with parental psychological utility when compared with negative and uncertain

Table 4. Multivariate linear regression of the educational utility subscale assessed at baseline, post-results disclosure, and 6m-post results disclosure

	Baseline (<i>n</i> = 908)		Post-results disclosure (<i>n</i> = 924)		6m Post-results disclosure (<i>n</i> = 828)	
	β (95% CI)	<i>p</i> value	β (95% CI)	<i>p</i> value	β (95% CI)	<i>p</i> value
Socioeconomic factors						
Education level						
HS/GED/post-HS training vs. <HS	1.08 (0.18, 1.97)	0.018	1.18 (0.10, 2.27)	0.032	1.60 (0.25, 2.94)	0.020
College graduate vs. <HS	0.43 (−0.64, 1.51)	0.430	0.23 (−1.08, 1.54)	0.726	0.85 (−0.80, 2.49)	0.311
College+ vs. <HS	0.18 (−1.00, 1.35)	0.769	0.39 (−1.05, 1.82)	0.598	0.16 (−1.64, 1.96)	0.862
Survey language						
Spanish vs. English	0.19 (−0.68, 1.06)	0.663	0.25 (−0.78, 1.28)	0.631	1.17 (−0.10, 2.45)	0.071
Insurance type						
Private vs. public	−0.28 (−0.99, 0.44)	0.447	−0.49 (−1.36, 0.38)	0.267	0.22 (−0.88, 1.32)	0.695
Healthcare distrust						
Values subscale	0.04 (−0.07, 0.14)	0.508	0.05 (−0.08, 0.19)	0.418	−0.01 (−0.17, 0.16)	0.965
Competence subscale	−0.15 (−0.29, −0.02)	0.029	−0.09 (−0.26, 0.08)	0.303	−0.14 (−0.35, 0.08)	0.206
Health Literacy Score	−0.01 (−0.11, 0.08)	0.779	−0.11 (−0.22, 0.01)	0.070	0.03 (−0.12, 0.17)	0.722
Clinical factors						
Case-level clinical interpretation						
Positive/likely positive vs. negative	–		3.02 (1.93, 4.11)	<0.001	0.62 (−0.74, 1.98)	0.372
Uncertain vs. negative	–		−0.59 (−1.41, 0.23)	0.159	−1.19 (−2.23, −0.16)	0.024
Positive/likely positive vs. uncertain ^a	–		3.61 (2.59, 4.62)	<0.001	1.81 (0.55, 3.07)	0.005
Previous genetic testing						
Yes vs. no	−0.01 (−0.62, 0.61)	0.982	0.12 (−0.63, 0.87)	0.757	−0.30 (−1.24, 0.65)	0.539

HS, high school; GED, General Education Development Test.

Bold entries signify significant results.

^aPost-hoc analyses were conducted using uncertain result as the reference variable for case-level clinical interpretation.

results, but only at the 6m post-results disclosure time point ($b = 1.17, p = 0.001$ and $b = 1.53, p < 0.001$, respectively). At post-results disclosure, there was no association seen for uncertain results when compared with negative results however, positive/likely positive results had higher parental psychological utility than uncertain results ($b = 1.84, p < 0.001$). We did not find an association between parental psychological utility and insurance type, healthcare distrust values subscale, and previous genetic testing.

Discussion

In this study, we explored perceptions of the personal utility of GT in a diverse cohort of participants whose children underwent GT through the NYCKidSeq program. Overall, parents/guardians identified high levels of personal utility for genetic testing. Parents'/guardians' views on personal utility were highest prior to undergoing testing and declined over time after learning the results, suggesting

that the actual utility of the genetic test results did not align with their initial expectations. Notably higher levels of personal utility were reported by those whose children received positive or likely positive results compared with negative and uncertain results, which is unsurprising given the potential clinical actionability and emotional relief of diagnostic results. In contrast to previous research, which has primarily comprised qualitative studies with small sample sizes and/or was typically conducted with highly educated, non-Hispanic White women, we examined personal utility in a socioeconomically, racially, ethnically, and linguistically diverse cohort.^{7,8,17–20,35} There were significant differences observed between H/L participants, who reported the highest levels of overall personal utility, compared with EA and AA participants. In addition, socio-demographic factors, including education, health literacy, and healthcare distrust, emerged as having a significant impact on personal utility. Understanding the differences in how population groups may perceive the utility of GT and the socioeconomic factors driving these differences

Table 5. Multivariate linear regression of the parental psychological subscale assessed at baseline, post-results disclosure, and 6m post-results disclosure

	Baseline (n = 917)		Post-results disclosure (n = 926)		6m Post-results disclosure (n = 831)	
	β (95% CI)	p value	β (95% CI)	p value	β (95% CI)	p value
Socioeconomic factors						
Education level						
HS/GED/post-HS training vs. <HS	0.56 (0.11, 1.00)	0.014	0.29 (-0.32, 0.89)	0.352	0.64 (-0.06, 1.34)	0.074
College graduate vs. <HS	0.19 (-0.35, 0.72)	0.497	-0.74 (-1.48, -0.01)	0.048	0.13 (-0.72, 0.99)	0.763
College+ vs. <HS	-0.33 (-0.92, 0.25)	0.261	-0.93 (-1.73, -0.12)	0.024	-0.61 (-1.54, 0.32)	0.199
Survey language						
Spanish vs. English	0.08 (-0.35, 0.50)	0.725	0.36 (-0.22, 0.93)	0.225	0.52 (-0.14, 1.18)	0.125
Insurance type						
Private vs. Public	-0.04 (-0.40, 0.31)	0.808	-0.34 (-0.82, 0.15)	0.175	-0.41 (-0.98, 0.16)	0.161
Health Literacy Score	-0.03 (-0.08, 0.02)	0.177	-0.07 (-0.14, -0.01)	0.033	-0.06 (-0.13, 0.02)	0.139
Healthcare distrust						
Values subscale	0.01 (-0.05, 0.06)	0.898	0.03 (-0.04, 0.11)	0.401	-0.08 (-0.12, 0.05)	0.397
Competence subscale	-0.06 (-0.13, 0.01)	0.029	-0.10 (-0.19, -0.01)	0.037	-0.08 (-0.19, 0.03)	0.139
Clinical factors						
Case-level clinical interpretation						
Positive/likely positive vs. negative	-		2.00 (1.39, 2.61)	<0.001	1.17 (0.47, 1.88)	0.001
Uncertain vs. negative	-		-0.16 (-0.30, 0.62)	0.491	-0.36 (-0.89, 0.18)	0.195
Positive/likely positive vs. uncertain ^a	-		1.84 (1.27, 2.41)	<0.001	1.52 (0.87, 2.19)	<0.001
Previous genetic testing						
Yes vs. no	-0.07 (-0.37, 0.24)	0.675	-0.28 (-0.70, 0.14)	0.191	-0.34 (-0.83, 0.15)	0.172

HS, high school; GED, General Education Development Test.

Bold entries signify significant results.

^aPost-hoc analyses were conducted using uncertain result as the reference variable for case-level clinical interpretation.

provides insights to inform provider communication and counseling strategies.

Identifying the various facets of personal utility (educational, psychological, and practical) allows for a deeper understanding of the motivations and values participants place on GT for their child. The high educational utility scores demonstrate that participants believe that a benefit of GT is its ability to provide information that may otherwise be unavailable. This finding is consistent with previous literature describing similar utilities, including providing families with sought-after information and satisfying their curiosity.^{5,11} Yet, educational utility identified through this work extends past the patient to include utility for other family members and the medical community as a whole. Likewise, identification of parent psychological utility allows us to expand on the previously defined “affective” domain of personal utility, which includes utilities of enhancing coping and mental preparation,⁵ to include enabling families to mentally prepare for their child’s

future health. Furthermore, participants found value in the practical utility of GT, which is centered on how GT can be used to improve the well-being of the child as well as the family. Although similar domains have been described previously as “pragmatic” or “behavioral” utilities, these primarily include actionable measures, such as informing long-term care, discussion with family about health conditions, and reproductive considerations.^{5,11,36} The practical utility we describe here also encompasses informing future plans for school or the child’s career and improving communication with family members. We also found that parents’ perceptions on the practical utility of GT may involve fewer tangible concepts, such as feeling more in control of the child’s life and health. As such, when contemplating GT, parents may not exclusively consider the impact of actionable recommendations on the child’s physical health but may also place importance on its emotional impact. Gaining these insights into the various aspects of personal utility helps to appreciate

how diverse families make decisions to undergo GT and how they adapt to their child's results. This deeper understanding is essential for healthcare professionals discussing genetic testing in pediatric settings.

We found that personal utility significantly declined from pre-test to post-test, and this decline continued even 6 months after results disclosure. This finding highlights the discrepancy between the anticipated and actualized personal utility of GT. It is consistent with existing qualitative research demonstrating parents' unmet expectations of GT, where parents often anticipated that the GT would conclusively diagnose their child's condition.^{13,18,19,37} Our study not only validates these findings in a diverse population, but underscores the importance of utilizing pre-test counseling strategies, as discussed in the previous literature, that foster realistic expectations. Moreover, our study enhances our appreciation of the necessity to address parental expectations during post-test counseling to effectively manage expectations throughout and beyond the GT process. In addition, our research builds on previous studies^{13,38,39} by delineating personal utility into practical, educational, and parental psychological domains. While there is often an emphasis on managing expectations relating to clinical utility, our findings offer insights for providers to tailor their pre- and post-test counseling to address these three domains of personal utility.

A noteworthy finding from this study is the interplay between trust, or distrust, in the competence of the healthcare system and perceptions of the personal utility of genetic testing. Low levels of trust were associated with low levels of personal utility prior to testing and after receiving results. This finding remained consistent for the overall summary score and for the parent psychological and practical utility domains. Trust has been suggested as an important factor related to attitudes toward receiving genetic and genomic results, specifically patients' trust in the ability of healthcare professionals to interpret and translate GT results into meaningful medical care.^{20,40} Notably, it was found that participants with less than a high school education reported higher levels of trust in the healthcare system and reported higher practical and parental psychological utility compared with those who were college educated. Perhaps participants with higher education levels perceived less utility in GT due to lower levels of trust and skepticism of the ability of the healthcare system to provide constructive and effective guidance based on the results. As this skepticism may not apply to the accuracy of the genetic test result itself, but rather its successful applicability,²⁰ this may explain why there was no association with educational utility and education level nor healthcare distrust. Studies have reported conflicting results on the relationship between educational attainment and healthcare distrust.^{41–43} Further research is needed to disentangle the potential contributors to personal utility of different levels of education and its relationship to distrust of the healthcare system.

When considering how diverse populations experience personal utility of pediatric GT, it is notable that H/L parents had higher personal utility scores than EA and AA parents. Two studies exploring attitudes of women toward prenatal genetic screening revealed that Spanish-speaking H/L women tended to express more trust in the advice given by their healthcare provider, were less inclined to question recommendations, and were more likely to favor a provider-driven approach to decision-making compared with non-H/L women.^{44,45} Therefore, to further explore factors that might be driving differences in personal utility across the three population groups, we performed a post-hoc stratified analysis that included additional sociodemographic factors, including parents' perceived and objective understanding of GT results, trust in the healthcare system, and mode of results. Although no differences in trust or mode of delivery were observed, interestingly, this analysis demonstrated across populations that higher perceived understanding was associated with greater personal utility, and higher levels of objective understanding were associated with lower personal utility scores. This suggests that perceptions of the understanding of GT results are a greater indicator of personal utility than actual understanding, suggesting that how individuals feel about their comprehension of the results may be more important to their sense of utility. However, no consistent pattern explained the initial observation of higher personal utility in H/L parents, indicating that personal utility from GT may be not solely dependent on the clinical result, but also shaped by perceptions of understanding in combination with other psychological, social, and systemic factors that vary among different populations which may not have been measured in this study.

Limitations of the study

There are limitations to this study that should be considered. The method in which participants received GT results depended on whether they were enrolled in the NYCKidSeq or the TeleKidSeq studies, which differed by use of telemedicine and/or screen sharing capabilities and by the use GUÍA, a digital platform designed to facilitate the communication of GT results. Although post-hoc analyses did not find a significant difference between personal utility levels in individuals who received results in-person vs. via telehealth, it is possible that these factors impacted personal utility. The PrU measure was adapted for participants of pediatric populations and has not been validated in this study population. Generalizability of the study findings may be limited as we recruited from one urban area, participants were predominantly mothers (potentially overshadowing the perspectives of fathers or legal guardians), and the majority of participants' children who underwent GT had a neurological phenotype.

Conclusions

Understanding the role of patient-centric factors, including personal utility, is critical for implementing

genetic testing in an evidence-based manner. This study, involving a diverse cohort of parents of children undergoing genetic testing, personal utility encompassed three key domains: practical, educational, and psychological utility. A range of factors influenced participant's views on personal utility across these domains. Most notable were time and the type of genetic result, as well as participants' education level, health literacy, and trust in the healthcare system. Additional research involving diverse patient populations in other clinical contexts may further elucidate factors impacting perceptions of the utility of GT to further inform pre- and post-test counseling for GT across a variety of populations and clinical settings. As the diversity of patients receiving GT expands, it is critical to understand the perspectives of those underrepresented populations who have been historically excluded from genomics research, and who may experience the utility of GT differently.

Data and code availability

The datasets used and/or analyzed during this study are available from the corresponding author on reasonable request.

Supplemental information

Supplemental information can be found online at <https://doi.org/10.1016/j.xhgg.2024.100321>.

Acknowledgments

Research reported in this article was supported by the National Human Genome Research Institute (NHGRI) and the National Institute on Minority Health and Health Disparities (NIMHD) of the National Institutes of Health under award no. U01HG009610. The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.

The authors thank all of the children, parents, and families who participated in this study. The authors also thank the New York Genome Center, Rady Children's Institute for Genomic Medicine, and the Sema4 laboratory; the members of the Mount Sinai Hospital Genomics Stakeholder Board; and referring physicians in the Mount Sinai and Montefiore Health Systems.

This work was supported in part through the computational and data resources and staff expertise provided by Scientific Computing and Data at the Icahn School of Medicine at Mount Sinai and supported by the Clinical and Translational Science Awards (CTSA) grant UL1TR004419 from the National Center for Advancing Translational Sciences.

Author contributions

L.J.B., B.D.G., C.R.H., G.A.D., J.M.G., M.P.W., N.S.A.-H., R.E.Z., and E.E.K. contributed to the conception and design of the study. P.N.M., S.A.S., J.A.O., K.B., K.M.G., M.S., M.A.R., M.D.B., and J.E.R. contributed to the acquisition of data. L.S. and B.J.I. performed the analysis. P.N.M., S.A.S., L.S., B.J.I., M.A.R., N.R.K., E.E.K., and C.R.H. contributed to the interpretation of data.

P.N.M., S.A.S., L.S., B.J.I., K.B., J.A.O., E.E.K., and C.R.H. drafted the manuscript. All authors read and approved the final manuscript.

Declaration of interests

N.S.A.-H. is currently employed by 23andMe, was previously employed by Regeneron Pharmaceuticals, received personal fees from Genentech, Allelica, and 23andMe, received research funding from Akcea, and serves as a scientific advisory board member for Allelica. E.E.K. received personal fees from Illumina, 23andMe, Allelica, and Regeneron Pharmaceuticals, received research funding from Allelica, and serves as a scientific advisory board member for Encompass, Bio, Overtone, and Galateo Bio.

Received: October 27, 2023

Accepted: June 21, 2024

Web resources

Find shortage areas by address: <https://data.hrsa.gov/tools/shortage-area/by-address>.

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Supplemental information

**Evaluating parental personal utility
of pediatric genetic and genomic testing
in a diverse, multilingual population**

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Supplemental Materials

Figure S1. Distrust in the competence of the healthcare system by education

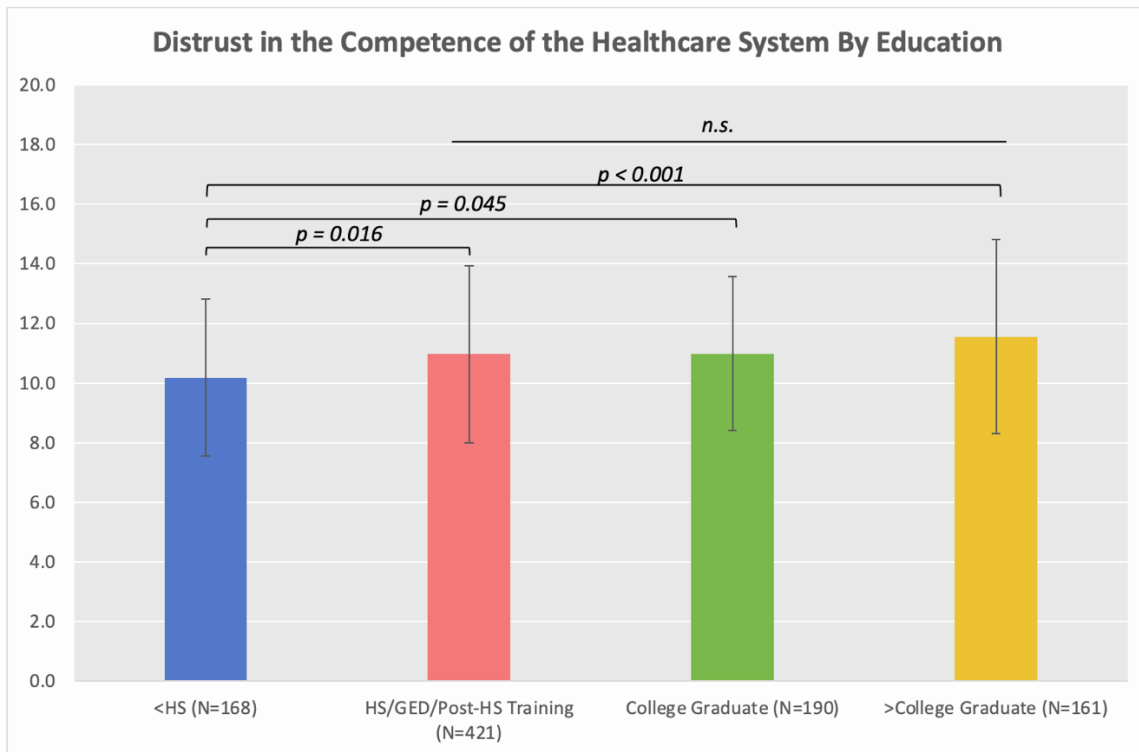


Figure S1. ANOVA and post hoc Tukey tests between highest level of education completed by parents/guardians and distrust in the competence of the healthcare system displayed above (($F(3,936) = 6.3, p = 0.001$). Displayed p-values reflect post-hoc Tukey results (n.s. = not significant). Those with less than a high school education scored significantly lower on the distrust scale compared to the other three education levels. No differences found between HS/GED/Post-HS Training, College Graduate and >College Graduate.

Abbreviations: HS: High school; GED: General Education Development Test

Figure S2. Personal utility summary score across three largest population groups

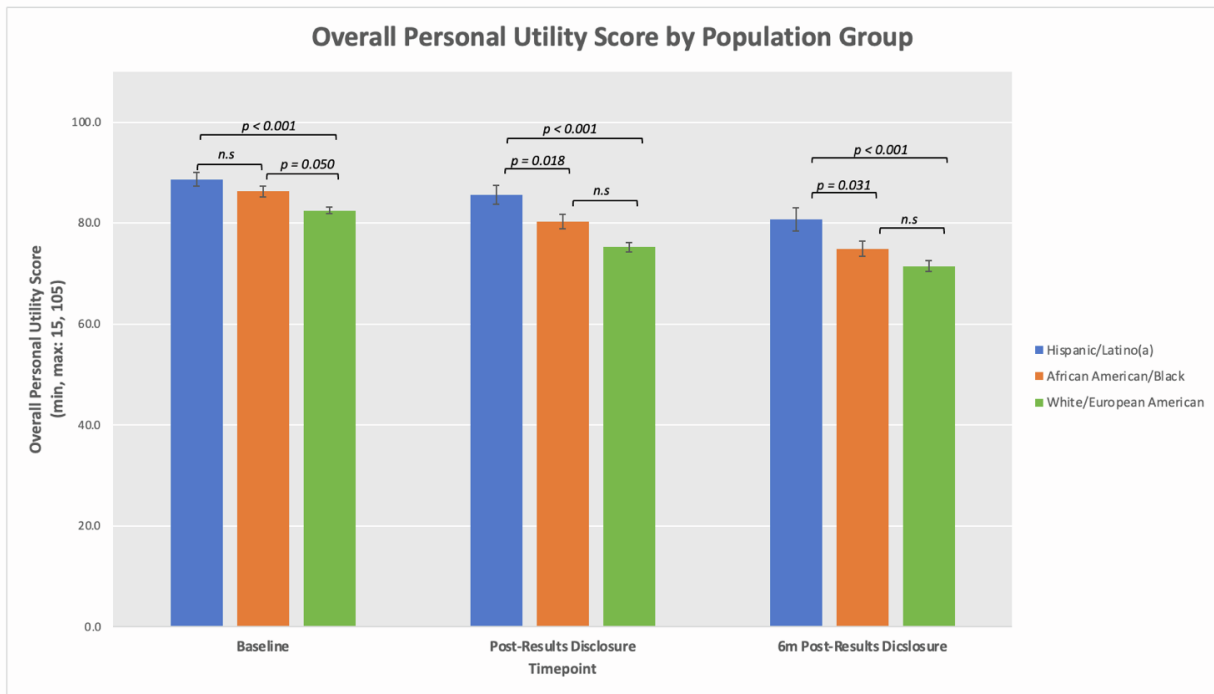


Figure S2. ANOVA and post-hoc Tukey tests between overall personal utility score and the three largest population groups (Hispanic/Latino(a) (H/L), African American/Black (AA) and European American (EA)) across three time points (baseline: $F(2,786) = 12.5$, $p < 0.001$; post-results disclosure: $F(2,785) = 19.5$, $p < 0.001$; 6m post-results disclosure: $F(2,713) = 12.0$, $p < 0.001$). Displayed p-values reflect post-hoc Tukey results (n.s. = not significant). Post-results disclosure, H/L participants had greater personal utility scores than EA and AA participants. At baseline, H/L participants had greater personal utility than EA participants, but H/L and AA participants did not significantly differ. At post-results disclosure and 6m post-results disclosure, no differences found between AA and EA.

Table S1. Factor analysis of the adapted pediatric Personal Utility Scale (PrU) measure (N=944)

Survey item ^a	Factor 1 ^b (<i>Practical Utility</i>)	Factor 2 ^c (<i>Informational Utility</i>)	Factor 3 ^d (<i>Parental Psychological Utility</i>)
1. Help with my child's life planning	0.78	0.29	0.20
2. Inform plans for my child's school or career	0.82	0.18	0.17
5. Help me or our family mentally prepare for the future	0.19	0.10	0.96
6. Help to better understand my child's health	0.17	0.12	0.96
7. Contribute to my child's self-knowledge	0.67	0.38	0.23
8. Help me cope with my child's health risks	0.74	0.39	0.25
9. Help me feel more in control of my child's health	0.78	0.33	0.20
10. Help me feel more in control of my child's life	0.77	0.30	0.19
11. Simply to provide information	0.31	0.75	0.08
12. Satisfy my curiosity about my child	0.43	0.65	0.15
13. Help my child use social programs, like resources and services	0.79	0.28	0.17
14. Improve communication with my family members	0.78	0.28	0.19
15. Feel good about helping the medical community	0.23	0.79	0.13
16. Feel good about having information for family members	0.45	0.64	0.17
17. Feel good about taking responsibility for my child's health	0.35	0.76	0.16

^a Original scale consisted of 17 items; however, only 15 items were used in this analysis, two items (items 3 and 4) were removed a priori since they were not applicable to the entire study cohort

^b Scale reliability for Practical Utility: Cronbach's $\alpha = 0.94$

^c Scale reliability for Informational Utility: Cronbach's $\alpha = 0.87$

^d Scale reliability for Parental Psychological Utility: Cronbach's $\alpha = 0.97$

Table S2. Outcomes assessed to evaluate personal utility

Measures	Survey question	Response options	Source	Time point		
				Baseline	Post-results disclosure	6m post-results disclosure
Outcome Measure						
Parent’s Expectations of Personal Utility of Genomic Results	<p>In this next section, we will ask you how useful you believe your child's test results will be in making future decisions. We know you have not yet received the results, but we want to know what you expect may happen when you do get them.</p> <p>Please indicate how useful you think your child's results will be for the following purposes:</p> <ol style="list-style-type: none"> 1. Help with my child’s life planning 2. Inform plans for my child’s school or career 5. Help me or our family mentally prepare for the future 6. Help to better understand my child’s health 7. Contribute to my child’s self-knowledge 8. Help me cope with my child’s health risks 9. Help me feel more in control of my child’s health 10. Help me feel more in control of my child’s life 11. Simply to provide information 12. Satisfy my curiosity about my child 13. Help my child use social programs, like resources and services 14. Improve communication with my family members 	<p>1, Not at all useful 2, A little useful 3, Somewhat useful 4, Neutral 5, Useful 6, Very useful 7, Extremely useful 98, Don’t Know</p>	<p>CSER measure adapted for pediatric populations from the Personal Utility Scale (PrU)(1)</p>	X	-	-

	<p>15. Feel good about helping the medical community</p> <p>16. Feel good about having information for family members</p> <p>17. Feel good about taking responsibility for my child's health</p>					
<p>Personal Utility of Genomic Results after Results Disclosure</p>	<p>In the next section, we will ask you questions about how useful your child's test results will be in making future decisions.</p> <p>Please indicate how useful you find the following outcomes of your child's test result:</p> <ol style="list-style-type: none"> 1. Help with my child's life planning 2. Inform plans for my child's school or career 5. Help me or our family mentally prepare for the future 6. Help to better understand my child's health 7. Contribute to my child's self-knowledge 8. Help me cope with my child's health risks 9. Help me feel more in control of my child's health 10. Help me feel more in control of my child's life 11. Simply to provide information 12. Satisfy my curiosity about my child 13. Help my child use social programs, like resources and services 14. Improve communication with my family members 15. Feel good about helping the medical community 16. Feel good about having information for family members 17. Feel good about taking responsibility for my child's health 	<p>1, Not at all useful 2, A little useful 3, Somewhat useful 4, Neutral 5, Useful 6, Very useful 7, Extremely useful 98, Refused</p>	<p>CSER measure adapted for pediatric populations from the Personal Utility Scale (PrU)(1)</p>	-	X	X

Table S3. Covariates and population characteristics included in the analysis

Provided as an excel due to size of table.

Table S4. Bivariate analyses of selected variables with Personal Utility Scale (PrU) summary score

Bivariate Analysis of Variables	N^a	PrU Summary Score, M(SD)	P-Value^b
Relationship to child			
Mother	1562/1782	79.4 (21.7)	0.27
Father	143/1782	76.4 (22.5)	
Legal Guardian	77/1782	78.3 (23.0)	
Health system			0.65
Montefiore Medical Center	605/1782	79.4 (23.6)	
Mount Sinai Health System	117/1782	79.0 (20.9)	
Previous genetics testing			0.008
No	1056/1782	80.3 (21.5)	
Yes	705/1782	77.5 (22.0)	
Self-reported race and ethnicity^c			<0.001
African American/Black	266/1782	77.8 (24.3)	
Hispanic/Latino(a)	830/1782	83.2 (20.4)	
White/European American	408/1782	73.5 (21.0)	
How was the survey administered?			0.98
Phone/Videoconference	1487/1782	79.1 (22.0)	
In-person	295/1782	79.1 (20.9)	
Education level			<0.001
< HS	321/1782	81.9 (22.2)	
HS Grad/GED/Post-HS Training	797/1782	82.5 (20.5)	
College graduate	355/1782	75.4 (21.9)	
> College graduate	303/1782	72.0 (22.5)	
Insurance Type			<0.001
Public	1150/1782	81.3 (21.3)	
Private	632/1782	75.2 (22.3)	
Case-level interpretation of genetic test result			<0.001
Positive/Likely Positive	315/1782	89.1 (18.2)	
Uncertain	906/1782	76.4 (21.6)	
Negative	561/1782	78.0 (22.6)	
Language of Survey during Immediate Timepoint			<0.001
Spanish	402/1782	85.0 (19.9)	
English	1380/1782	77.4 (22.1)	
Language of Survey during 6m Timepoint			<0.001
Spanish	1327/1782	84.6 (20.6)	
English	379/1782	77.7 (22.0)	
Brief Health Literacy Score	1782/1782	79.1 (21.8)	<0.001
Healthcare Distrust Values Subscale Score	1780/1782	79.1 (21.8)	<0.001
Healthcare Distrust Competence Subscale Score	1780/1782	79.1 (21.8)	<0.001

^a N reflects number of responses from both timepoints: post-results disclosure and 6 months post-results disclosure. Variations in total N (N=1782) reflect missing values in the original variables.

^b T-tests conducted for variables with 2 categories, one-way analysis of variance conducted for variables with 3 or more variables, Pearson's correlation conducted for continuous variables.

^c Only the three largest race and ethnicity groups used in the bivariate analysis: African American/Black, Hispanic/Latino(a) and White/European American.

Abbreviations: HS: High school; GED: General Education Development Test

Table S5. Multivariate linear regression of the personal utility summary score assessed at post-results disclosure using measures of understanding and mode of result delivery

	Full Sample (N=912) ^a		EA Only (N=212)		AA Only (N=137)		H/L Only (N=417)	
	β (95% CI)	p-value	β (95% CI)	p-value	β (95% CI)	p-value	β (95% CI)	p-value
Measures of Understanding								
Perceived Understanding								
Level 2 vs Level 1	6.84 (2.22, 11.46)	0.004	19.47 (5.81, 33.12)	0.005	10.39 (-2.34, 23.11)	0.109	7.52 (1.33, 13.70)	0.017
Level 3 vs Level 1	10.17 (5.81, 14.56)	<0.001	22.18 (9.27, 35.10)	0.001	18.18 (5.19, 31.16)	0.007	9.26 (3.58, 14.94)	0.001
Objective Understanding Summary Score	-2.27 (-3.46, -1.08)	<0.001	-2.83 (-5.92, 0.27)	0.073	-5.01 (-8.67, -1.34)	0.008	-1.72 (-3.35, -0.09)	0.038
Mode of Result Disclosure								
In Person vs Telehealth	-0.97 (-4.45, 2.50)	0.583	1.81 (-5.94, 9.56)	0.646	4.40 (-6.04, 14.83)	0.405	0.59 (-4.30, 5.47)	0.814
Socioeconomic Factors								
Education Level								
HS/GED/Post-HS training vs <HS	1.59 (-2.13, 5.31)	0.402	-1.73 (-22.11, 18.65)	0.867	4.78 (-5.89, 15.46)	0.376	3.21 (-1.21, 7.63)	0.154
College Graduate vs <HS	-4.52 (-9.01, -0.02)	0.049	-7.04 (-27.77, 13.69)	0.504	5.73 (-7.95, 19.42)	0.408	-1.46 (-7.39, 4.46)	0.628
College+ vs <HS	-5.14 (-10.07, -0.22)	0.041	-1.49 (-22.22, 19.23)	0.887	-5.61 (-20.87, 9.65)	0.468	-5.4 (-14.09, 3.29)	0.223
Survey Language								
Spanish vs English	1.65 (-1.95, 5.26)	0.368	- ^b	-	9.33 (-20.99, 39.65)	0.543	-0.38 (-4.72, 3.97)	0.864
Insurance Type								
Private vs Public	-2.09 (-5.09, 0.90)	0.171	5.10 (-1.29, 11.49)	0.117	-1.62 (-9.88, 6.63)	0.698	-4.99 (-9.84, -0.14)	0.044
Health Literacy Score	-0.48 (-0.90, -0.05)	0.027	-0.91 (-2.13, 0.30)	0.141	-1.19 (-2.57, 0.18)	0.088	-0.28 (-0.81, 0.25)	0.295
Healthcare Distrust								
Values Subscale	0.23 (-0.22, 0.68)	0.319	0.89 (-0.130- 1.92)	0.087	-0.68 (-1.96, 0.59)	0.290	0.36 (-0.28, 0.99)	0.273
Competence Subscale	-0.76 (-1.33, -0.18)	0.010	-1.51 (-2.73, -0.29)	0.016	-0.52 (-2.14, 1.10)	0.528	-0.67 (-1.52, 0.19)	0.126
Clinical Factors								
Case-Level Clinical Interpretation								
Positive/Likely Positive vs Negative	14.62 (10.85, 18.39)	<0.001	15.12 (6.89, 23.35)	<0.001	21.29 (8.67, 33.91)	0.001	13.89 (8.70, 19.07)	<0.001
Uncertain vs Negative	-0.66 (-3.63, 2.31)	0.663	0.08 (-6.69, 6.85)	0.981	-4.99 (-13.95, 3.97)	0.272	1.70 (-2.48, 5.88)	0.425
Previous Genetic Testing								
Yes vs No	-2.12 (-4.74, 0.50)	0.112	-3.5 (-9.15, 2.16)	0.225	6.56 (-1.79, 14.91)	0.122	-4.29 (-8.06, -0.53)	0.026

^a While not shown, all models controlled for the genetic counselor who returned the results.

^b Language omitted from model due to collinearity.

Abbreviations: EA: White or European American; AA: Black or African American; H/L= Hispanic/Latino(a)

Table S6. Multivariate linear regression of the personal utility summary score assessed at 6m post-results disclosure using measures of understanding and mode of result delivery

	Full Sample (N=800) ^a		EA Only (N=183)		AA Only (N=116)		H/L Only (N=380)	
	β (95% CI)	p-value	β (95% CI)	p-value	β (95% CI)	p-value	β (95% CI)	p-value
Measures of Understanding								
Perceived Understanding								
Level 2 vs Level 1	6.91 (2.71, 11.11)	0.001	1.19 (-9.87, 12.24)	0.833	6.65 (-5.46, 18.76)	0.278	4.36 (-1.41, 10.13)	0.138
Level 3 vs Level 1	8.00 (3.94, 12.06)	<0.001	5.33 (-5.54, 16.21)	0.334	5.35 (-7.13, 17.82)	0.397	6.77 (1.24, 12.30)	0.017
Objective Understanding Summary Score	-1.19 (-2.43, 0.04)	0.058	-0.37 (-3.31, 2.58)	0.805	2.53 (-1.82, 6.87)	0.250	-1.21 (-2.88, 0.45)	0.153
Mode of Result Disclosure								
In Person vs Telehealth	1.17 (-2.96, 5.29)	0.579	1.40 (-7.03, 9.84)	0.743	-1.32 (-15.04, 12.41)	0.849	2.95 (-2.90, 8.81)	0.322
Socioeconomic Factors								
Education Level								
HS/GED/Post-HS training vs <HS	0.73 (-3.66, 5.13)	0.744	-1.31 (-23.02, 20.41)	0.906	14.41 (-0.09, 28.91)	0.051	-0.88 (-6.08, 4.32)	0.740
College Graduate vs <HS	-2.85 (-8.18, 2.48)	0.294	0.95 (-21.17, 23.08)	0.932	6.94 (-10.81, 24.69)	0.439	-5.43 (-12.49, 1.63)	0.131
College+ vs <HS	-7.33 (-13.18, -1.48)	0.014	0.16 (-21.94, 22.25)	0.989	-6.55 (-25.76, 12.66)	0.500	-9.75 (-19.71, 0.22)	0.055
Survey Language								
Spanish vs English	2.44 (-1.81, 6.69)	0.260	-. ^b	-	-. ^b	-	-0.18 (-5.15, 4.79)	0.942
Insurance Type								
Private vs Public	-3.09 (-6.62, 0.44)	0.086	-2.05 (-9.285, 5.179)	0.576	0.57 (-9.80, 10.93)	0.914	-4.64 (-10.18, 0.90)	0.101
Health Literacy Score	-0.12 (-0.61, 0.37)	0.632	-1.11 (-2.48, 0.29)	0.119	0.3 (-1.41, 2.01)	0.725	0.12 (-0.493, 0.732)	0.701
Healthcare Distrust								
Values Subscale	-0.27 (-0.80, 0.26)	0.316	0.78 (-0.34, 1.91)	0.171	-1.23 (-2.86, 0.41)	0.139	-0.12 (-0.86, 0.63)	0.755
Competence Subscale	-0.64 (-1.32, 0.04)	0.067	-1.36 (-2.75, 0.03)	0.056	-0.27 (-2.30, 1.77)	0.796	-0.91 (-1.91, 0.09)	0.075
Clinical Factors								
Case-Level Clinical Interpretation								
Positive/Likely Positive vs Negative	7.18 (2.82, 11.54)	0.001	9.78 (0.75, 18.80)	0.034	1.72 (-13.81, 17.25)	0.827	5.57 (-0.47, 11.60)	0.071
Uncertain vs Negative	-3.48 (-6.93, -0.02)	0.049	-3.36 (-11.31, 4.59)	0.405	-11.97 (-23.16, -0.78)	0.036	-1.6 (-6.45, 3.26)	0.518
Previous Genetic Testing								
Yes vs No	-2.32 (-5.39, 0.74)	0.137	-9.24 (-15.66, -2.82)	0.005	-1.21 (-11.27, 8.86)	0.812	0.77 (-3.63, 5.16)	0.732

^a While not shown, all models controlled for the genetic counselor who returned the results.

^b Language omitted from model due to collinearity.

Abbreviations: EA: White or European American; AA: Black or African American; H/L= Hispanic/Latino(a)

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