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Perceived Benefits, Risks, and Utility of Newborn Genomic Sequencing in the BabySeq Project

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Abstract

BACKGROUND AND OBJECTIVES: There is interest in applying genomic sequencing (GS) to newborns' clinical care. Here we explore parents' and clinicians' attitudes toward and perceptions of the risks, benefits, and utility of newborn GS compared with newborn screening (NBS) prior to receiving study results.

METHODS: The BabySeq Project is a randomized controlled trial used to explore the impact of integrating GS into the clinical care of newborns. Parents ($n = 493$) of enrolled infants ($n = 309$) and clinicians ($n = 144$) completed a baseline survey at enrollment. We examined between-group

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This trial has been registered at www.clinicaltrials.gov (identifier NCT02422511).

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differences in perceived utility and attitudes toward NBS and GS. Open-ended responses about risks and benefits of each technology were categorized by theme.

RESULTS: The majority of parents (71%) and clinicians (51%) agreed that there are health benefits of GS, although parents and clinicians agreed more that there are risks associated with GS (35%, 70%) than with NBS (19%, 39%; all $P < .05$). Parents perceived more benefit and less risk of GS than did clinicians. Clinicians endorsed concerns about privacy and discrimination related to genomic information more strongly than did parents, and parents anticipated benefits of GS that clinicians did not.

CONCLUSIONS: Parents and clinicians are less confident in GS than NBS, but parents perceive a more favorable risk/benefit ratio of GS than do clinicians. Clinicians should be aware that parents' optimism may stem from their perceived benefits beyond clinical utility.

Nearly all infants born in the United States receive state-mandated newborn screening (NBS) at birth. Fifty years after being implemented to screen newborns for phenylketonuria, allowing early intervention to avoid resulting disabilities, NBS programs have expanded to now screen for >30 conditions that present in the newborn period for which there is early treatment or management.^{1,2} The program is largely hailed as a success and is credited with improving the health of newborns by preventing or mitigating the symptoms of certain conditions.^{3,4}

Continued technological advancements in genomic sequencing (GS), alongside its increased affordability, have generated growing interest in applying GS to the clinical care of newborns.^{5,6} Some have even suggested that every infant will be sequenced at birth in the not-too-distant future.^{7,8} Others have noted the inevitability of introducing GS into or alongside current NBS programs.^{9,10} GS of newborns has the potential to provide comprehensive information of clinical and personal utility to clinicians and families.^{6,11–13} Advocates of GS expect its utility to surpass that of NBS because GS could shorten the time to diagnose diseases¹⁴ by interrogating thousands of disease-causing variants at once¹⁵ and detecting conditions that NBS currently cannot.¹¹ Additionally, GS can identify the risk of future disease thus creating opportunities for early interventions to prevent or prepare for future health problems.^{2,4,15} Critics of GS remind us that despite its promise, GS is unlikely to replace all screening, because biochemical tests like tandem mass spectrometry are superior at detecting some disorders.⁹ Limitations of GS also include our incomplete knowledge about control elements and the identification of variants of uncertain significance.

Despite the potential for GS to enrich current NBS programs, the increased amount of information about an individual that can be generated and the implications of this information for family members' own health or family planning decisions has led to considerable debate.^{2,4,15} For example, the ethical implications of the ability of GS to identify risk for later-onset disorders in newborns raises questions about the impact on a child's right to an open future.^{4,16–20} Professional guidelines urge caution with regard to the use of GS in children, advise against returning information associated with adult-onset conditions to parents of children,^{16,21,22} recommend testing only individual or targeted panels of genes when such testing will suffice,²¹ and generally discourage sequencing

healthy children.^{21,22} In addition to the potential for ambiguity in the interpretation of results,²³ the psychological and psychosocial impact of receiving GS information on families is as of yet unknown.²⁴

Research has revealed that many parents express hypothetical interest in receiving their newborn's GS information,^{12,25} yet little is known about the perspectives of those who might actually receive GS information about their newborns or those who provide medical care for them. In this article, we explore attitudes toward and perceptions of the risks, benefits, and utility of GS compared with standard NBS among clinicians and parents participating in a randomized controlled trial of GS in newborns.

METHODS

Study Design and Participants

The BabySeq Project is a randomized clinical trial exploring the medical, behavioral, and economic impact of integrating GS into the clinical care of newborns. The study design has been described in detail elsewhere.²⁶ To summarize, we are enrolling newborns, their parents, and their clinicians into 2 cohorts: a healthy newborn cohort from Brigham and Women's Hospital (BWH) Well Baby Nursery and an ICU newborn cohort from ICUs at BWH, Boston Children's Hospital, and Massachusetts General Hospital. Within each cohort, half of the families are randomly assigned to modified standard of care (standard NBS report and an in-depth family history report), and half are randomly assigned to modified standard of care plus a type of GS (whole-exome sequencing). Families in the GS arm receive a genome report with results about monogenic and recessive carrier variants in genes associated with conditions that present or for which there are interventions during the childhood period, as well as a set of highly penetrant, actionable adult-onset conditions and pharmacogenomics variants. For newborn participants who have or develop a clinical presentation that may have a genetic etiology, families may receive results from an indication-focused sequence analysis. Genome reports are uploaded into the newborn's medical record at the associated hospital and sent to clinicians involved in their care. The Partners Human Research Committee, which serves as the review board for BWH, Boston Children's Hospital Office of Clinical Investigations, and Baylor College of Medicine's Institutional Review Board approved this study.

The infant's parents and clinicians, including primary care physicians and pediatric subspecialists, complete multiple surveys throughout the study. In this report, we focus on parents' and clinicians' baseline attitudes toward and perceptions of the benefits, risks, and utility of GS compared with NBS. Both groups completed baseline surveys after enrollment into the project before they knew their randomization status and before any results were returned.

Measures

Ten items were used to assess parents' and clinicians' attitudes about obtaining informed consent versus mandating testing, perceptions of the risks and benefits, and concerns about privacy and discrimination of NBS and GS. Response options to these questions were on a

5-point Likert scale anchored by “strongly disagree” to “strongly agree.” Parents were asked to rate the importance of having each type of information (NBS and GS) about their newborn now and in 10 years from now on a 5-point scale anchored by “not at all important” (1) and “very important” (5) to measure their perceived utility. To evaluate clinicians’ perceived utility, they were asked to rate the usefulness of each type of technology at this time and in 10 years for identifying conditions in their patients, managing their patients’ care, and predicting their patients’ future risk of disease. Responses were on a 10-point scale anchored by “not at all useful” (1) to “extremely useful” (10).

Data Analysis

Descriptive statistics were calculated to characterize parents’ and clinicians’ demographic characteristics and responses to survey items. We compared attitudes toward NBS versus GS among parents and clinicians using χ^2 and Wilcoxon signed rank tests. Paired *t* tests were used to assess whether there were differences in perceived current and future utility (defined as 10 years from now) of NBS versus GS among parents and clinicians. Wilcoxon signed rank tests were used to explore differences between parents’ and clinicians’ perceived current and future utility of each technology (NBS versus GS). To achieve this, we first calculated the difference in the mean current and future utility scores for each technology of parents and clinicians. All statistical calculations were performed using SPSS statistical software (IBM SPSS Statistics, IBM Corporation).

Parents and clinicians who agreed or strongly agreed that there were risks or benefits of each technology were then asked to specify those risks and benefits via open-ended responses. We analyzed these open-ended responses qualitatively using thematic content analysis. We identified themes of responses using an inductive approach, with which 3 coders, who were overseen by a fourth coder with qualitative expertise (S.P.), coded all responses independently and then discussed any discrepancies until consensus was reached.

RESULTS

Participant Characteristics

We enrolled 493 parents of 309 newborns and 144 clinicians. Parents’ and clinicians’ characteristics are presented in Tables 1 and 2. Parents who completed surveys were 50% women, the majority were college graduates or higher (88%), and 68% were non-Hispanic white. The majority of clinicians were white (82%), and 90% had no specific genetics training.

Attitudes and Perceived Risks and Benefits

Summary statistics of parents’ and clinicians’ attitudes toward NBS and GS are presented in Table 3. The vast majority of parents (93%) and all clinicians (100%) agreed that all newborns should receive NBS, but only 33% of parents and 8% of clinicians felt that all newborns should receive GS ($P < .001$). Parents agreed more than clinicians that parental informed consent should be required for NBS (57%, 31%, respectively; $\chi^2 P = .001$), but parents and clinicians felt similarly that parental informed consent should be required for GS (86%, 93%; $\chi^2 P = .058$).

These attitudes are in line with their views toward the risks and benefits of these technologies, as both groups agreed more that there are health benefits associated with NBS (82% of parents, 97% of clinicians) than with GS (71%, 51%; all $P < .05$), although parents had a more favorable view of the health benefits of GS than clinicians ($\chi^2 P < .001$).

Likewise, parents and clinicians agreed more that there are risks associated with GS (35%, 70%) than with NBS (19%, 39%; all $P < .05$), although clinicians agreed significantly more than parents that there are risks of both technologies (GS $\chi^2 P < .001$; NBS $\chi^2 P < .001$).

Seventy-two percent of clinicians agreed that parents should be concerned about discrimination based on their child's genomic information, but only 53% of parents reported being concerned about discrimination ($\chi^2 P < .001$). Likewise, 83% of clinicians agreed that parents should be concerned about the privacy of their child's genomic information, whereas only 65% of parents expressed concern about it ($\chi^2 P < .001$). In open-ended responses (Table 4), parents and clinicians both cited the potential for discrimination as more of a risk of GS than of NBS. They also cited the potential to receive uncertain or unwanted results, such as results associated with untreatable or later-onset disorders, as more of a risk of GS than of NBS, with clinicians in particular noting the risks associated with the potential for uncertain results with GS (eg, variants of unknown significance, issues of penetrance). Both parents and clinicians identified psychological distress as a risk of both NBS and GS, describing the possibility of parents feeling anxious or depressed in response to results from either technology.

In terms of benefit, parents and clinicians saw diagnosis and/or identification of risk and early intervention as the main benefits of both NBS and GS, citing the potential to mitigate or prevent symptoms associated with a broad range of disorders, although the potential to identify the risk of future conditions was a much stronger theme in responses about GS than about NBS. Other benefits identified through open-ended responses included family planning and testing, the ability to prepare for health problems that may arise (exclusive of clinical intervention, including mental preparation or parental education), and valuing information solely for the sake of knowledge (ie, "knowledge is power"), although the ability to prepare and the value of information were largely absent from clinicians' responses.

Perceived Utility

As shown in Fig 1, parents rated the importance of having NBS information higher than having GS information now but rated the 2 types of information as equally important 10 years from now. When we examined the differences between parents' current and future utility perceptions, we found they thought that GS information would be significantly more important in 10 years compared with now ($P < .001$) and that the importance of having NBS information would decrease over the next 10 years ($P < .001$).

Clinicians similarly responded that NBS was more useful than GS at this time for identifying conditions in their patients (Table 5; $P < .001$) and managing their patients' care ($P < .001$) but felt NBS and GS were equally useful at this time for predicting their patients' future risk of disease ($P > .05$). In terms of the future utility of these technologies, clinicians indicated that NBS will still be more useful than GS for identifying conditions in their

patients ($P < .05$) but felt that GS will be more useful than NBS for predicting their patients' future risk of disease ($P < .001$). We also examined differences between clinicians' perception of current and future utility and found that they similarly expect GS to be more useful in 10 years compared with now (all 3 purposes $P < .001$). Although clinicians responded that NBS would increase in utility over time for managing their patients' care and predicting their patients' future risk of disease, they anticipated the utility of GS to increase more than the utility of NBS over the next 10 years for all 3 purposes (Wilcoxon signed rank test $P < .001$).

We found no differences in any attitude items between types of clinicians (neonatologists versus pediatricians), nor between cohorts of parents (Well Baby Nursery versus ICU).

DISCUSSION

Overall, parents and clinicians viewed NBS more favorably than GS. Parents and clinicians were less confident in GS compared with NBS, but the majority of both parents and clinicians agreed that there are health benefits of newborn GS. Parents perceived more benefit and less risk of GS than did clinicians, with clinicians viewing the difference in risks and benefits between the 2 technologies as more disparate than parents did. Clinicians may be viewing GS as riskier on the basis of their concerns about privacy issues and discrimination related to genomic information, which they endorsed more strongly than parents. As such, parents seem to perceive a more favorable benefit/risk ratio of GS than clinicians.

Parents' and clinicians' open-ended responses describing the risks and benefits of GS provide some additional insight into the way they perceive GS. Although both parents and clinicians most often identified psychological distress as a risk of GS, clinicians also often cited the uncertainty associated with genomic information and many times linked that to parents' distress, suggesting that GS could cause unnecessary parental distress by causing parents to worry about health problems that may never arise or that the child is not actually at risk for developing. Clinicians also more often identified the possibility of receiving unwanted results, such as results associated with later-onset conditions. These concerns recall the potential harms of GS noted in the literature, such as impinging on the child's right to an open future^{4,16–20} or disrupting family dynamics, like parent-child bonding.²⁴

Parents also anticipated benefits that clinicians did not, including the ability to prepare and the benefit of knowledge for its own sake. These results echo other studies that have revealed that parents find their children's genomic information useful for a broad variety of reasons, including psychological and pragmatic reasons,^{13,27} which traditionally fall outside of the realm of what is considered clinical utility, because these benefits do not lead to a change of clinical management.²⁸

Professional guidelines and recommendations about sequencing in children emphasize that the benefits of the test should be weighed against the risks. The definition of utility of genomic technologies, however, is currently an area of debate, with some arguing that a broader spectrum of utility should be recognized with regard to genomic technologies^{29,30}

and others emphasizing that consumers' and patients' perception of use does not necessarily mean the information actually has utility.³¹ Our results reveal that both parents and clinicians identify benefits of GS that are not typically covered under the umbrella of what is considered "clinical utility," such as family planning and testing, the intrinsic value of information, and the ability to prepare for the future. Additionally, parents and clinicians view the risks and benefits of GS differently. Underscored in these findings is the importance of further exploring the utility of genomic technologies during the newborn period, broadly defined. In addition, clinicians should be aware that parents might have a more optimistic view of GS and should be prepared to discuss with parents the risks, benefits, and goals of using GS to help set realistic expectations.

Our results should be considered within the limitations of our study. First, we are reporting on the perspectives of parents who have chosen to participate in the BabySeq Project. As such, it is unsurprising that they hold a relatively optimistic view of GS; parents who declined to participate may have a less favorable attitude. In addition, the majority of respondents were highly educated and white. Thus, these views may not be generalizable to a larger population of parents. Additionally, we have reported attitudes toward these 2 technologies at baseline, and although a small number of parents of ICU infants may have already received NBS results, most families had not, and none had yet received their study results, nor had any clinicians received any study results for their patients. Follow-up surveys are used to query parents' and clinicians' attitudes toward these 2 technologies again after they have received results (3 months postdisclosure for parents and end of study for clinicians); analysis of these surveys will be used to determine whether receiving study results impacts their attitudes. Future analyses will also explore whether there is concordance between parents' attitudes, because disagreements in perspectives could affect clinical practice. Finally, parents who considered enrollment in the study participated in an ~1-hour education and consent session with a genetic counselor, during which they were educated about the risks and benefits of GS, which may have had an effect on their attitudes.

CONCLUSIONS

As we continue to introduce GS into the clinical care of newborns, there is a need to understand parents' and clinicians' attitudes toward and perceived utility of GS at this life stage. Understanding how parents and clinicians perceive GS, especially in comparison with the successful and longstanding NBS program in the United States, can provide insight into how parents and clinicians may view the integration of GS and how they may respond to GS results. Our results reveal that parents and clinicians are less confident in GS than NBS, but parents are more optimistic about GS than are clinicians. Clinicians should be aware that parents' optimism may stem from a broader range of perceived benefits that are outside of what is generally considered clinical benefit, and as such, they may need to help set realistic expectations for GS information. Further research into the utility of GS information for infants and their families will help clinicians and parents weigh the risks and benefits of using GS in the newborn period.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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ABBREVIATIONS

BWH	Brigham and Women's Hospital
GS	genomic sequencing
NBS	newborn screening

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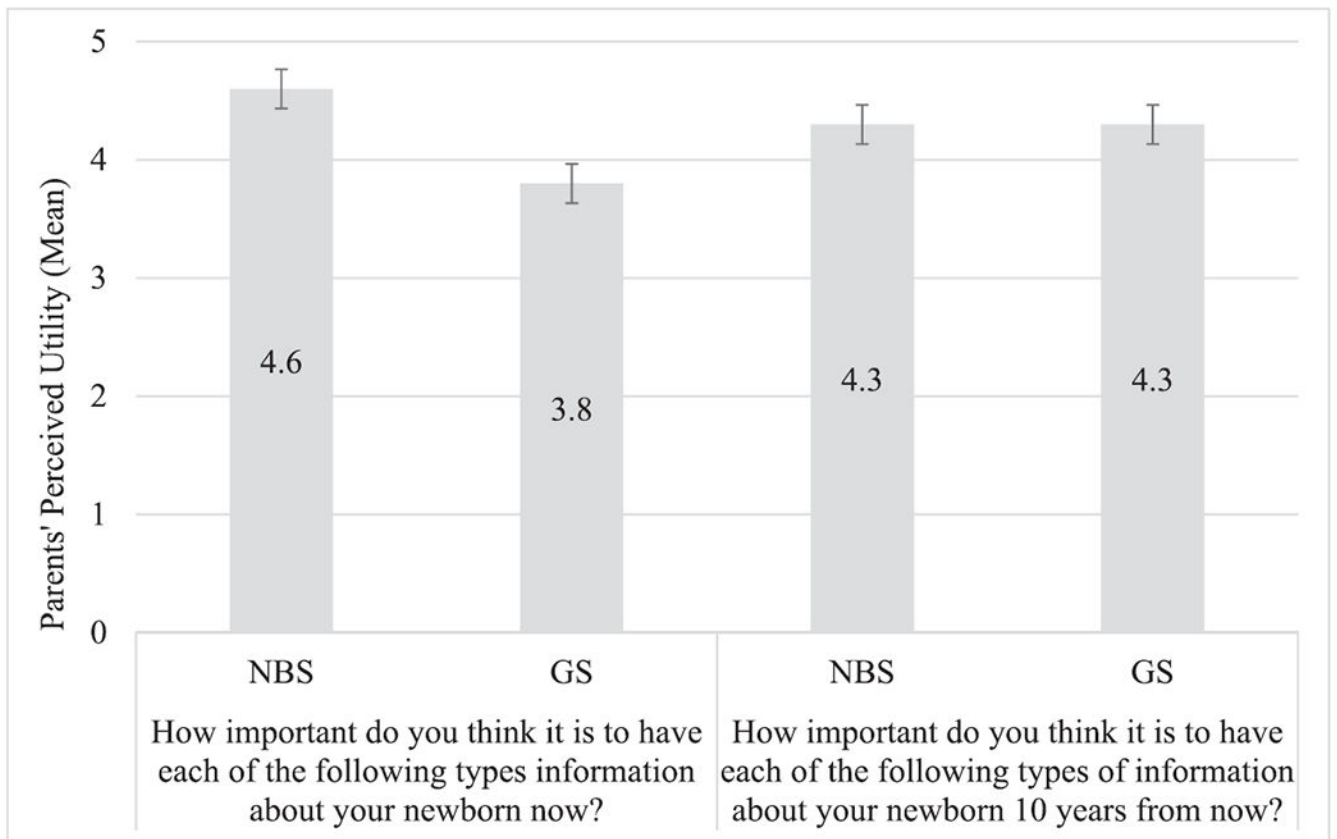


FIGURE 1.

Parents' perceived utility. Parents' perceived utility is measured on an importance scale that ranges from 1 to 5 in which 1 indicates no importance and 5 indicates high importance. Error bars represent SEs.

TABLE 1

Parent Characteristics

Characteristic, <i>n</i> (%) Unless Otherwise Noted	<i>N</i> = 493
<i>Age</i> ^{<i>a</i>}	
Mean in <i>y</i> (SD)	35.9 (±4.8)
<i>Sex</i>	
Male	247 (50%)
Female	246 (50%)
<i>Race and/or ethnicity</i>	
Hispanic or Latino	32 (6%)
Non-Hispanic white	335 (68%)
Non-Hispanic other ^{<i>b</i>}	126 (26%)
<i>Education</i>	
Did not graduate from college	59 (12%)
College graduate or higher	434 (88%)
<i>Annual household income</i> ^{<i>c</i>}	
\$99 999	97 (20%)
\$100 000	386 (80%)

^{*a*} *n* = 476.

^{*b*} Non-Hispanic other includes African American, Asian American, and other.

^{*c*} *n* = 483.

TABLE 2

Clinician Characteristics

Characteristic, n (%) Unless Otherwise Noted	N = 148
<i>Age</i> ^a	
Mean in y (SD)	46.0 (±13.8)
<i>Sex</i>	
Male	50 (35%)
Female	98 (66%)
<i>Race and/or ethnicity</i> ^b	
Hispanic or Latino	5 (4%)
Non-Hispanic white	118 (82%)
Non-Hispanic other ^c	21 (15%)
<i>Years in practice</i> ^d	
1–10	35 (25%)
11–20	53 (37%)
21	51 (37%)
<i>Genetics training</i> ^e	
No	127 (90%)
Yes	14 (10%)
<i>Clinician type</i>	
Neonatologist	53 (36%)
Pediatrician	92 (62%)
Clinician other ^f	3 (2%)

^a
n = 133.

^b
n = 144.

^cNon-Hispanic other includes African American, Asian American, and other.

^d
n = 139.

^e
n = 141.

Clinician other includes cardiologist and family medicine practitioner.

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TABLE 3

Parents' and Clinicians' Attitudes

	Parents' Attitudes				Clinicians' Attitudes			
	N	Disagree, %	Neither Agree nor Disagree, %	Agree, %	N	Disagree, %	Neither Agree nor Disagree, %	Agree, %
Every newborn should receive								
NBS	494	1	6	93	146	—	—	100
GS	494	10	57	33	146	42	50	8
Parental informed consent should be required for								
NBS	493	22	21	57	147	54	15	31
GS	492	3	11	86	147	1	5	93
There are health benefits associated with								
NBS	493	2	16	82	145	1	2	97
GS	494	2	27	71	147	5	44	51
There are risks associated with								
NBS	490	50	31	19	144	48	13	39
GS	493	28	37	35	148	4	26	70
Concerned about the privacy of child's genomic Information ^{a,b}	493	17	18	65	145	4	13	83
Concerned child will be discriminated against on the basis of genomic information ^{c,d}	490	22	25	53	147	8	20	72

Response options were on a scale ranging from strongly disagree (1) to strongly agree (5). Scores 1 and 2 were combined to create the disagree category. Scores 4 and 5 were combined to create the agree category. —, not applicable.

^aParent survey question: I am concerned about the privacy of my child's genomic information.

^bClinician survey question: My patients' parents should be concerned about the privacy of their child's genomic information.

^cParent survey question: I am concerned that my child will be discriminated against on the basis of his or her genomic information.

^dClinician survey question: My patients' parents should be concerned that their child will be discriminated against on the basis of his or her genomic information.

TABLE 4
Themes of Benefits and Risks in Parents’ and Clinicians’ Open-ended Responses

Theme	Definition
Benefits	
Ability to prepare	Awareness of health problems that may arise can aid in preparation and planning, exclusive of clinical intervention
Diagnosis and/or identification of risk and early intervention	Early diagnosis, identification of risk for diseases; prevention, early treatment, and intervention
Family planning and testing	Carrier status, family testing
Value of information	Knowledge for its own sake; “knowledge is power”
Risks	
Analytic validity	Test sensitivity, false-positives, and false-negatives
Uncertain results	Issues of penetrance, variants of unknown significance
Impact on family	Family members treating child differently on the basis of results
Privacy and discrimination	Risks of privacy of results; child could face genetic and insurance discrimination
Psychological distress	Anxiety, worry, stress for patient or family
Unwanted results	Receiving unwanted or too much information; receiving results associated with later-onset conditions

Clinicians' Perceived Use

TABLE 5

	How Useful Do You Think NBS Is at This Time		How Useful Do You Think NBS Will Be in 10 y		How Useful Do You Think GS Is at This Time		How Useful Do You Think GS Will Be in 10 y	
	N	Mean (SD)	N	Mean (SD)	N	Mean (SD)	N	Mean (SD)
For identifying conditions in your patients	148	8.9 (±1.4)	148	8.9 (±1.5)	148	6.1 (±1.9)	146	8.4 (±1.6)
For managing your patients' care	148	7.8 (±2.4)	147	8.2 (±2.1)	147	5.6 (±2.2)	146	7.8 (±1.9)
For predicting your patients' future risk of disease	146	6.3 (±2.9)	148	7.4 (±2.5)	146	6.2 (±2.1)	147	8.3 (±1.6)

Clinicians' perceived utility is measured on a usefulness scale that ranges from 1 to 10 in which 1 indicates no usefulness and 10 indicates high usefulness.