

Attitudes toward genomic medicine among US reproductive-age adults and parents enrolled in genomic screening research

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Abstract

Background

Genomic sequencing (GS) may soon be integrated into population-based newborn and pediatric screening programs. Successful GS implementation will depend in part on public attitudes towards genomic medicine, yet little is known about the extent to which parents who enroll their infants in GS research reflect the general public in terms of attitudes toward genomic medicine, or how parental experience with GS impacts these attitudes.

Methods

We assessed attitudes toward genomic medicine among a representative sample of US reproductive-age adults using the Genomic Orientation (GO) scale, which consists of optimism and pessimism subscales. Within the BabySeq Project, a multi-site randomized controlled trial of GS, we administered longitudinal parental surveys which included the GO scale at the time of enrollment and six months after result disclosure. We compared mean optimism and pessimism scores between the public and BabySeq samples. To evaluate the impact of GS trial participation, we used linear regression with fixed effect for baseline scores to evaluate BabySeq parent scores over time by trial arm (control vs. GS), genomic result type (Mendelian disease risk identified vs. no reportable findings), and parent characteristics.

Results

Compared with US reproductive-age adults ($n = 1,021$), BabySeq parent-participants who completed the GO scale ($n = 411$; 91.2% female, 68.1% underrepresented race or ethnicity) were more optimistic (5.8 vs 3.3, $p < 0.001$) and less pessimistic (-0.8 vs 0.4, $p < 0.001$) toward genomic medicine at enrollment. Over the course of the trial, parents became both less optimistic (by mean - 1.3 points, $p < 0.001$) and less pessimistic (by mean - 1.3 points, $p < 0.001$). Parents with a high school diploma or less had mean six month post-disclosure optimism scores that were 1.1 points lower than parents with more than a high school diploma ($p = 0.025$). Differences in scores by trial arm or genomic result type were not observed.

Conclusion

Parents who enrolled their infants in BabySeq were more optimistic and less pessimistic about the future of genomic medicine than US reproductive-age adults. While optimism and pessimism decreased amongst BabySeq parents over time, experience with GS did not impact attitudes toward genomic medicine. These findings have implications for the scale-up of GS research studies into population-based screening programs.

Trial Registration:

The BabySeq Project is a registered clinical trial (clinicaltrials.gov identifier NCT05161169).

Background

There is interest in using genomic sequencing (GS) as a screening tool (genomic screening) to identify risk for rare genetic disease early in life. Genomic screening during the newborn period or infancy provides the opportunity for surveillance, early diagnosis, and timely treatment of hundreds of genetic diseases. Numerous newborn genomic screening pilot and implementation projects are underway both across the United States (US) and around the world (1). Within the US, the GUARDIAN study (2) and EarlyCheck study (3) incorporate genomic sequencing as a complement to traditional newborn screening (NBS) using residual dried bloodspot samples, and the second iteration of the BabySeq Project incorporates genomic sequencing into well-baby pediatric care within the first year of life (4). Additionally, the recently federally funded BRIDGES-NBS initiative, which will assess the feasibility of integrating genomic sequencing into NBS in seven state public health programs across the US (5), as well as the state-funded Sunshine Genetics pilot program in Florida (6), represent substantial steps toward implementation of population-based genomic screening for newborns.

The successful implementation of population-based genomic screening programs will depend upon parental acceptability and uptake, influenced by public attitudes towards genomic medicine. A survey conducted in 2012 of a population-representative sample of parents in the US found that a majority of parents were at least somewhat interested in genomic screening being implemented as a public health program (7). Another US study conducted in 2012 and 2013 found that a majority of new parents at a single hospital that were queried within 48 hours of birth were at least somewhat interested in genomic testing for their newborn (8). However, the high hypothetical interest in genomic screening seen in these studies did not ultimately translate to high enrollment rates in the first iteration of the BabySeq Project, a genomic screening trial that enrolled newborns from 2015 to 2017, with only 6.7% of approached families consenting to enroll in the study (9). Given the substantial amount of time since these studies were conducted and the proliferation of newborn genomic screening pilot projects in recent years, an updated understanding of public attitudes towards genomic medicine is needed.

While research studies such as the first iteration of the BabySeq Project have surveyed parents to understand psychosocial impacts and attitudes toward genomic screening and NBS among parents who chose to enroll in the trial (10, 11), these participants were not representative of the US population in terms of demographic characteristics (9), and as they were among some of the first to volunteer to participate in this type of research, they may also have held more positive attitudes toward genomic medicine overall. The second iteration of the BabySeq Project enrolled a more demographically diverse cohort (4); however, it has yet to be established whether and to what extent BabySeq participants' views of genomic medicine reflect those of US adults who may soon be eligible to enroll their infants in a population-based genomic screening program. We sought to understand attitudes towards genomic medicine among the diverse sample of parents who enrolled their infants in the second iteration of the BabySeq Project. Specifically, we evaluated whether parents who enrolled their infants in this study differed from the a representative sample of reproductive-age US adults in terms of optimism and

pessimism toward genomic medicine, as well as whether and how study participation impacted their attitudes toward genomic medicine.

Methods

Sample and survey administration

This study assessed and compared attitudes toward genomic medicine among a US population-representative sample of reproductive-age adults (age 18–49 years) and parents who enrolled their infant in a genomic screening research study. We assessed attitudes toward genomic medicine using the Genomic Orientation (GO) scale, a validated measure of optimism and pessimism toward genomic medicine (12, 13). Within the scale completion instructions, genomic medicine is defined as the use of genome sequencing in health care. The GO scale asks participants to rate the likelihood that genomic medicine will have various impacts within the next five years in 26 items across six conceptual domains: cost, medical benefit, health disparities, individual control, inevitability, and discovery. The 10-item GO optimism subscale and the 16-item GO pessimism subscale are scored from –10 (strong disagreement with the subscale construct) to 10 (strong agreement with the subscale construct). In the public survey, we also included a global item in which participants were asked to indicate the statement that best described their views about the potential of genomic medicine, taking all things into consideration.

A brief survey including the GO scale was fielded in English to a US population-representative sample of reproductive-age adults (age 18–49) by Verasight, a survey research firm that maintains a panel of participants recruited via random address-based sampling, random person-to-person text messaging, and dynamic online targeting (14). All participants included in the final sample provided consent to participate and passed all data quality assurance checks, which included confirmation that responses corresponded with a US IP address and there were no duplicate or non-human respondents, as well as removal of respondents who failed straight-lining checks and that completed the survey in less than 30% of the median completion time.

Our sample of parents participating in genomic screening research comes from the second iteration of the BabySeq Project, which was a multi-site randomized controlled trial of genomic screening of infants in pediatric clinical care that began enrolling in January 2023. Families were randomized to either the control arm, which received a family history report, or to the GS arm, which received GS plus a family history report. Study participation was available in English and Spanish. Study methods are reported in greater detail elsewhere (4). Parents who enrolled in BabySeq were invited to complete longitudinal surveys to assess parent-reported psychosocial, health, and health care utilization outcomes. These surveys included the GO scale. One parent of each infant was invited to complete surveys at the time of trial enrollment, at the time after result disclosure, and six months after results disclosure. Parents completed the baseline survey before randomization, and thus baseline responses reflect attitudes of parents who elected to enroll their infant in a genomic screening program.

Statistical analysis

In this analysis, we analyzed survey participants' responses to the GO scale. The GO scale was scored according to published scoring instructions, which includes within-person mean imputation. Across both the public and BabySeq parent samples, we included only participants who skipped one or fewer items per subscale (across both survey timepoints for BabySeq parents). Only BabySeq parents who completed both baseline and six-month post-disclosure surveys between January 2023 (when recruitment began) and June 2026 (when the vast majority of trial participants would have been at least six months post-disclosure) were included in analyses.

To evaluate whether attitudes towards genomic medicine among US reproductive-age adults differed by current parental status, we used two-sample t-tests to compare subscale scores stratified by whether participants endorsed currently being the parent or guardian of a child under 18 years of age. To evaluate whether attitudes toward genomic medicine among US reproductive-age adults differed by planned parental status, we used ANOVA to compare subscale scores stratified by whether participants endorsed planning to have a child within the next five years (yes, no, uncertain).

To evaluate whether BabySeq parents' attitudes toward genomic medicine differed from those of US reproductive age adults, we used two-sample t-tests to compare BabySeq parents' baseline optimism and pessimism subscale scores with those from the population sample. To evaluate whether BabySeq parents' attitudes toward genomic medicine differed from parents in the general population, we also conducted these analyses comparing BabySeq parents with the subgroup of the population sample who reported currently being the parent or guardian of a child under 18 years old.

To evaluate changes in BabySeq parents' attitudes over the course of trial participation, we used multivariable linear regression models with six-month post-disclosure subscale score as the outcome variable and baseline subscale score, parental race and ethnicity, household income, parental educational attainment, and preferred language as independent variables. Participants who left these items blank or responded "prefer not to answer" were not included in models, with the exception of household income, where "prefer not to answer" responses were included due to a sizable proportion (15.5%) of participants selecting this response option.

To evaluate whether any change in BabySeq parents' attitudes over time was associated with the experience of undergoing genomic screening and/or receiving a genomic finding, we included indicator variables for trial arm (control vs. GS) and genomic result type (Mendelian disease risk [MDR] identified vs. no reportable finding) in the model. We tested interaction terms between baseline score and trial arm and baseline score and genomic result type, neither of which were statistically significant and thus were not included in the final models.

Results

US reproductive-age adults

The public survey was fielded from June 4 through June 15, 2026. Out of a total of 1,027 responses, 6 were omitted due to item-level missingness and 1,021 were included in our analysis. Among these, 454 (44.6%) participants reported being a parent or guardian of a child under 18 years of age. Additionally, 214 (21.0%) reported planning to have a child within the next five years, while 175 (17.2%) were unsure. At the time they received the survey, slightly more than half of participants reported being not at all familiar with genomic medicine (n = 521, 51.0%), while a sizable minority reported being somewhat familiar (n = 431, 42.2%) and a small minority reported being very familiar (n = 69, 6.8%). After completing the GO scale, a majority of participants endorsed a view that, taking all things into consideration, genomic medicine will do a lot of good but also will create many problems (n = 535, 52.5%). Smaller proportions endorsed a view that genomic medicine will do a lot of good and will not create many problems (n = 325, 31.9%), genomic medicine will not do a lot of good but also will not create many problems either (n = 117, 11.5%), or genomic medicine will not do a lot of good and will create many problems (n = 43, 4.2%). Additional demographic information on the population representative sample can be found in Table 1, and their responses to individual GO scale items can be found in Supplementary Table 1. Responses to individual GO scale items stratified by self-reported parental status can be found in Supplementary Table 2.

Table 1
Population representative sample demographic
characteristics (n = 1,021)

Demographic characteristic	n (%)
Age	
Mean (SD)	35.35 (8.84)
Gender	
Male	477 (46.8%)
Female	533 (52.3%)
Other	10 (1.0%)
Race	
Black or African-American	151 (14.8%)
Pacific Islander	4 (0.4%)
Asian or Asian-American	89 (8.7%)
White	651 (63.8%)
Native American or American Indian	19 (1.9%)
Mixed race	74 (7.3%)
Some other race	32 (3.1%)
Hispanic ethnicity	
Yes	207 (20.3%)
No	814 (79.7%)
Current parent of a child under age 18	
Yes	454 (44.6%)
No	565 (55.4%)
Planning to have a child within 5 years	
Yes	214 (21.0%)
No	631 (61.9%)
Not sure	175 (17.2%)
Education	
Some high school or less	23 (2.3%)

Demographic characteristic	n (%)
Age	
High school graduate or GED	264 (25.9%)
Some college, no degree	174 (17.0%)
2-year or associate degree	102 (10.0%)
4-year or bachelor degree	290 (28.4%)
Post-graduate degree	168 (16.5%)
Income	
Less than \$15,000	86 (8.4%)
\$15,000 to under \$50,000	132 (13.0%)
\$50,000 to under \$75,000	274 (26.9%)
\$75,000 to under \$100,000	187 (18.4%)
\$100,000 to under \$150,000	175 (17.2%)
\$150,000 to under \$200,000	75 (7.4%)
More than \$200,000	90 (8.8%)
Census region	
Northeast	186 (18.3%)
Midwest	175 (17.2%)
South	379 (37.3%)
West	275 (27.1%)

The population-based sample was modestly optimistic about genomic medicine, with a mean optimism subscale score of 3.3. We observed differences in optimism towards genomic medicine by current parental status: participants who reported being the parent of a child under 18 years old were more optimistic than those who were not parents (mean 3.7 vs 3.0, $p = 0.002$). We also observed differences by planned parental status: those who reported planning to have a child within the next 5 years were more optimistic about genomic medicine (mean 3.8) than those who were not planning to (mean 3.1) or were unsure (mean 3.5, $p = 0.017$).

The general population was slightly pessimistic about genomic medicine, with a mean pessimism subscale score of 0.4. We observed differences in pessimism towards genomic medicine by current parental status: participants who reported being the parent of a child under 18 years old were more pessimistic than those who were not parents (mean 0.7 vs 0.2, $p = 0.011$). However, no differences in

pessimism by planned parental status were observed (mean 0.5 planning, 0.4 not planning, 0.3 unsure; $p = 0.853$).

BabySeq parents

In the BabySeq sample, out of 476 participants eligible for the six-month post-disclosure survey (those who attended their results disclosure session and were sent a survey six months after it), our analysis included GO scale scores for 411 participants (86.3%) at both the baseline and six-month post-disclosure survey timepoints. BabySeq parents were majority female (91.2%) and of underrepresented race or ethnicity (68.1%), defined here as Black or Hispanic. Detailed demographic characteristics are available in Table 2. BabySeq parent responses to individual GO scale items at baseline can be found in Supplementary Table 3 and at six-months post-disclosure in Supplementary Table 4.

Table 2
BabySeq parent demographic characteristics (total n = 411)

Parent sex	Control arm (n = 214)	Sequencing arm (n = 197)
Female	195 (91.1%)	180 (91.4%)
Male	19 (8.9%)	17 (8.6%)
Parent race and ethnicity		
Asian	10 (4.7%)	12 (6.1%)
Black	86 (40.2%)	80 (40.6%)
White	22 (10.3%)	20 (10.2%)
Hispanic/Latino	57 (26.6%)	57 (28.9%)
Other (incl. two or more races)	39 (18.2%)	28 (14.2%)
Parent education		
Less than high school	5 (2.3%)	5 (2.5%)
Some high school	19 (8.9%)	15 (7.6%)
High school graduate	55 (25.7%)	50 (25.4%)
Some post-high school training	32 (15.0%)	34 (17.3%)
Associate (2-year) college degree or certificate	35 (16.4%)	24 (12.2%)
Bachelor's degree	36 (16.8%)	44 (22.3%)
Graduate or professional degree	25 (11.7%)	22 (11.2%)
Prefer not to answer	5 (2.3%)	1 (0.5%)
No response	2 (0.9%)	2 (1.0%)
Household income		
Less than \$20,000	65 (30.4%)	57 (28.9%)
\$20,000 to \$39,999	43 (20.1%)	38 (19.3%)
\$40,000 to \$59,999	23 (10.7%)	26 (13.2%)
\$60,000 to \$79,999	14 (6.5%)	11 (5.6%)
\$80,000 to \$99,999	11 (5.1%)	9 (4.6%)
\$100,000 to \$139,999	9 (4.2%)	13 (6.6%)

Parent sex	Control arm (n = 214)	Sequencing arm (n = 197)
\$140,000 or more	13 (6.1%)	12 (6.1%)
Prefer not to answer	33 (15.4%)	30 (15.2%)
No response	3 (1.4%)	1 (0.5%)
Preferred language		
English	189 (88.3%)	173 (87.8%)
Spanish	19 (8.9%)	18 (9.1%)
No response	6 (2.8%)	6 (3.0%)
Household WIC/SNAP use		
Yes	142 (66.4%)	128 (65.0%)
No	57 (26.6%)	60 (30.5%)
I don't know	3 (1.4%)	3 (1.5%)
Prefer not to answer	10 (4.7%)	4 (2.0%)
No response	2 (0.9%)	2 (1.0%)

Attitudes toward genomic medicine among US reproductive-age adults compared with BabySeq parents

At baseline, BabySeq parents were more optimistic about genomic medicine compared with the general population (5.8 vs 3.3, respectively, $p < 0.001$; Fig. 1a). Similarly, BabySeq parents were less pessimistic about genomic medicine at baseline compared with the general population (-0.8 vs 0.4, respectively, $p < 0.001$; Fig. 1a). This finding held true in the subgroup analysis of parents in the general population: BabySeq parents were more optimistic at baseline than parents in the general population (5.8 vs 3.7, respectively, $p < 0.001$) and were less pessimistic at baseline than the parents in the general population (-0.8 vs 0.7, respectively, $p < 0.001$).

At six months post-disclosure, BabySeq parents remained more optimistic compared with the general population (mean 4.5 vs 3.3, respectively, $p < 0.001$; Fig. 1a). Similarly, BabySeq parents remained less pessimistic at six months post-disclosure compared with the general population (mean - 2.1 vs 0.4, respectively, $p < 0.001$; Fig. 1a). In subgroup analyses, BabySeq parents remained more optimistic at six months post-disclosure compared with parents in the general population (mean 4.5 vs 3.7, respectively, $p < 0.001$) and remained less pessimistic at six months post-disclosure compared with parents in the general population (mean - 2.1 vs 0.7, respectively, $p < 0.001$).

Impact of BabySeq participation on parents' attitudes toward genomic medicine over time

Overall, BabySeq parents became less optimistic over time (Fig. 1a & 1b), with a mean optimism score of 5.8 at baseline and 4.5 at six months post-disclosure ($p < 0.001$, mean change score -1.3 (SD 4.2)).

BabySeq parents also became less pessimistic over time (Fig. 1a & 1b), with a mean pessimism score of -0.8 at baseline and -2.1 at six months post-disclosure ($p < 0.001$, mean change score -1.3 (SD 4.6)).

Impact of exposure to genomic sequencing and result type on BabySeq parents' attitudes toward genomic medicine

In multivariable models, mean optimism scores at six months post-disclosure did not differ between trial arms (difference = 0.36; $p = 0.366$; Table 3). Parents with a high school diploma or less had mean six-month post-disclosure optimism scores that were 1.11 points lower than parents with more than a high school diploma (95% CI: -2.08 to -0.14 ; $p = 0.025$; Table 3). Parental race and ethnicity, household income, and preferred language did not have a significant effect on optimism scores (all $p > 0.06$; Table 3).

We found no effect of trial arm on mean pessimism scores at six-months post-disclosure (difference = -0.08 ; $p = 0.832$; Table 4). We found no effect of parental race and ethnicity, household income, parental education level, or preferred language (all $p > 0.06$; Table 4) on pessimism scores at six-months post-disclosure.

Table 3
Linear regression of optimism by trial arm (n = 387)

Six months post-disclosure optimism subscale score	coefficient	95% confidence interval	p-value
Baseline optimism	0.61	0.46 to 0.76	< 0.001
Trial arm			
Control arm	Ref.	Ref.	Ref.
GS arm	0.36	-0.42 to 1.14	0.366
Parent race/ethnicity			
Black	Ref.	Ref.	Ref.
Asian	-0.85	-2.41 to 0.72	0.288
White	0.49	-0.66 to 1.65	0.402
Hispanic/Latino	-1.06	-2.19 to 0.06	0.065
Other	0.57	-0.44 to 1.58	0.269
Household income			
Less than \$40,000	Ref.	Ref.	Ref.
\$40,000 or more	-0.09	-1.03 to 0.85	0.854
Prefer not to answer	0.07	-1.16 to 1.29	0.915
Parent education			
High school or less	-1.11	-2.08 to -0.14	0.025
More than high school	Ref.	Ref.	Ref.
Preferred language			
English	Ref.	Ref.	Ref.
Spanish	-1.21	-3.13 to 0.72	0.219

Table 4
Linear regression of pessimism by trial arm (n = 387)

Six months post-disclosure pessimism subscale score	coefficient	95% confidence interval	p-value
Baseline pessimism	0.43	0.31 to 0.55	< 0.001
Trial arm			
Control arm	Ref.	Ref.	Ref.
GS arm	-0.08	-0.87 to 0.70	0.832
Parent race/ethnicity			
Black	Ref.	Ref.	Ref.
Asian	0.4	-1.16 to 1.95	0.615
White	0.77	-0.62 to 2.16	0.278
Hispanic/Latino	-0.52	-1.58 to 0.54	0.337
Other	0.59	-0.60 to 1.78	0.327
Household income			
Less than \$40,000	Ref.	Ref.	Ref.
\$40,000 or more	0.88	-0.06 to 1.82	0.065
Prefer not to answer	1.31	-0.11 to 2.73	0.071
Parent education			
High school or less	0.13	-0.79 to 1.06	0.774
More than high school	Ref.	Ref.	Ref.
Preferred language			
English	Ref.	Ref.	Ref.
Spanish	-1.13	-3.01 to 0.76	0.24

Within the GS arm (n = 197), 23 (11.7%) had MDR findings. We found no effect of GS result type (MDR identified vs. no reportable finding) on optimism scores at six months post-disclosure (difference = 0.33; p = 0.658; Supplementary Table 4), although the effect of parental education level remained significant (difference = -1.69; 95% CI: -3.13 to -0.26; p = 0.021; Supplementary Table 5). We found no effect of parental race and ethnicity, household income, or preferred language (all p > 0.07; Supplementary Table 5) on optimism scores at six months post-disclosure.

Similarly, we found no effect of GS result type on pessimism scores at six months post-disclosure (difference = 1.25; $p = 0.133$; Supplementary Table 6). We found no effect of parental race and ethnicity, household income, parental education level, or preferred language (all $p > 0.06$; Supplementary Table 6) on pessimism scores at six months post-disclosure.

Discussion

In this study of attitudes toward genomic medicine among a representative sample of US reproductive-age adults and a diverse cohort of parents who enrolled their infant into a GS research study, we found that parents who elect to participate in GS research were generally more optimistic and less pessimistic toward genomic medicine than the general population. This was true at baseline and throughout the study. BabySeq parents became both less optimistic and less pessimistic as time went on. Those changes in attitudes did not appear to be a result of exposure to genomic medicine, as we found no effect of receiving GS results (i.e., being randomized to the sequencing arm), nor of receiving GS results indicating that their infant was at heightened risk of genetic disease.

Within the representative sample of US reproductive-age adults, we found that participants who were parents of children under the age of 18 years were both more optimistic and more pessimistic toward genomic medicine than those who were not parents. While substantial work has been done both within the US and internationally to examine public and parental views on GS as a screening tool for newborns and infants (15), there appears to be limited information available regarding the impact of parental status on attitudes towards genomics in general. Importantly, future work examining this topic should not only be limited to current parental status; within the representative sample of US reproductive-age adults, we found that participants who were planning to have a child within the next five years were more optimistic about genomic medicine than those who were not planning to have a child or were unsure. Within the context of infant GS program implementation, an emphasis should be placed on understanding the attitudes of those who are planning to have a child soon regardless of their current parental status; eliciting these perspectives could help shape the design of the enrollment process for future GS programs.

At the time of GS trial enrollment, BabySeq parents may be overestimating the capabilities and potential benefits of genomic medicine and underestimating the limitations of genomic medicine. The timing of the surveys and recency bias may contribute to this finding, as at the time of the baseline survey, parents had just spoken with a clinical research coordinator to provide consent, which included hearing about potential genomic results and actionability. At the time of the six-month post-disclosure survey, parents typically had not interacted with a study team member and may not have considered genomics nor their child's results for at least six months.

Both optimism and pessimism about genomic medicine decreased over time among BabySeq parents. It is possible that the experience of participating in a GS trial, regardless of trial arm or genomic result type received, enlightened parents to the limitations of genomic medicine that they may not have been aware

of at the time of trial enrollment. Such limitations include the limited impact that knowing one's genetic information has on healthcare for most people; in pediatric and adult populations alike, only a minority of those who undergo GS as a screening tool have actionable findings (2, 3, 16). A potential newfound understanding of the relatively limited impact of genomic information on most people's health could have decreased BabySeq parents' optimism toward genomic medicine. Parents may have perceived that if genomic medicine is less likely to do good than they initially thought, then it is also less likely to do harm than they initially thought, thereby decreasing pessimism as well. Future work, such as interviews or focus groups with parents who participated in the trial, could further elucidate these findings.

We found that parents with lower educational attainment had lower optimism toward genomic medicine at six months post-disclosure. Recent studies have shown that individuals with greater educational attainment have increased disease-related genetic literacy (17), awareness of precision medicine (18), and awareness of genetic testing (19). Education-related differences in genomic attitudes are likely to be correlated with health literacy and genomic knowledge; one study of patients with inflammatory bowel disease found that genetic literacy was correlated with education, and that those with higher genetic literacy had greater perceived utility of genetic testing when presented with a hypothetical genetic testing scenario (20). There are myriad factors, such as result comprehension and genetic literacy, that were not included in our analysis that may have further clarified the nature of our findings related to educational attainment.

Our finding that parents who enroll their infants in a GS study differ from the general population in their attitudes toward genomic medicine should be taken into account when considering the generalizability of other survey-based GS pilot study findings. In particular, other psychosocial findings generated from newborn and infant GS studies should be contextualized with the understanding that participants who enroll in these studies appear to have more favorable views of genomic medicine than the general public. Care must be taken to avoid designing a population-based GS program with the assumption that parents who enroll their infants in GS pilot studies have attitudes that are representative of parents as a whole, as doing so may risk overestimating uptake of and overall enthusiasm for a population-based GS program.

This study has some strengths and limitations. Strengths include the diversity of our cohort of parents who enrolled in a GS trial, the majority of whom are from demographic groups underrepresented in genomics research. Limitations include the imbalance in exposure to and familiarity with GS and genomic medicine between BabySeq parents and the population sample; BabySeq parents had more exposure to the concepts of GS and genomic medicine due to receiving education during the study's informed consent process, but the population sample reported limited knowledge of genomic medicine and received a brief written overview of GS but no formal educational materials.

Conclusions

Parents who enrolled their child in BabySeq had more optimistic and less pessimistic views on genomic medicine than the general public, parents became less optimistic and less pessimistic over the course of their participation in the trial, and there were no differences by randomization arm or genomic result type. Parents with a high school diploma or less were less optimistic at six months post-disclosure. Findings have implications for understanding the extent to which other survey-based GS study findings may hold true when GS is implemented as a population-based initiative and may help inform consent considerations and educational resources for large-scale population GS efforts, including transparency about the benefits and limitations of genomic screening.

Abbreviations

GS
genomic sequencing
NBS
newborn screening
GO scale
Genomic Orientation scale
MDR
Mendelian disease risk

Declarations

Ethics approval and consent to participate:

The BabySeq Project was approved by the Boston Children's Hospital IRB. The population survey was approved by the Harvard Pilgrim Health Care IRB (study #2412447). Participants in the BabySeq Project provided informed consent to participate in the trial. Participants in the population survey provided consent by proceeding to the survey after reading a consent page.

Consent for publication:

Not applicable.

Competing Interests

ALM is a paid advisor for Nurture Genomics. RCG has received compensation for advising Allelica, Atria, Fabric, Genome Web, and Genomic Life and is a cofounder of Genome Medical and Nurture Genomics. HSS has received consulting income from Illumina unrelated to this work.

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Author Contribution

MRH, KDC, and HSS contributed to conceptualization, methodology, and data curation. MRH and HSS conducted the formal analysis and created data visualizations. MRH, BZ, PA, KDC, and HSS conducted the investigation. MRH, PA, and HSS prepared the original draft. All authors (MRH, BZ, PA, SP, ALM, IAH, RCG, KDC, and HSS) contributed to review and editing. SP, ALM, IAH, RCG, KDC, and HSS provided supervision. IAH and RCG acquired funding for the study.

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Data Availability

Deidentified data are available upon request to the corresponding author.

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Figures

Genomic Orientation Scale

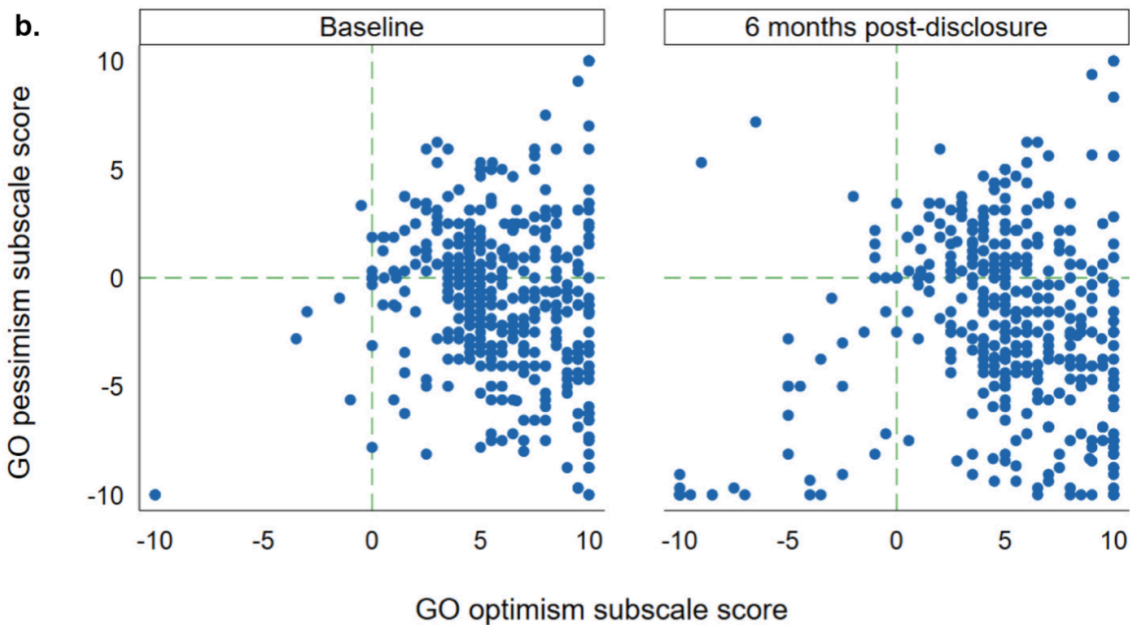
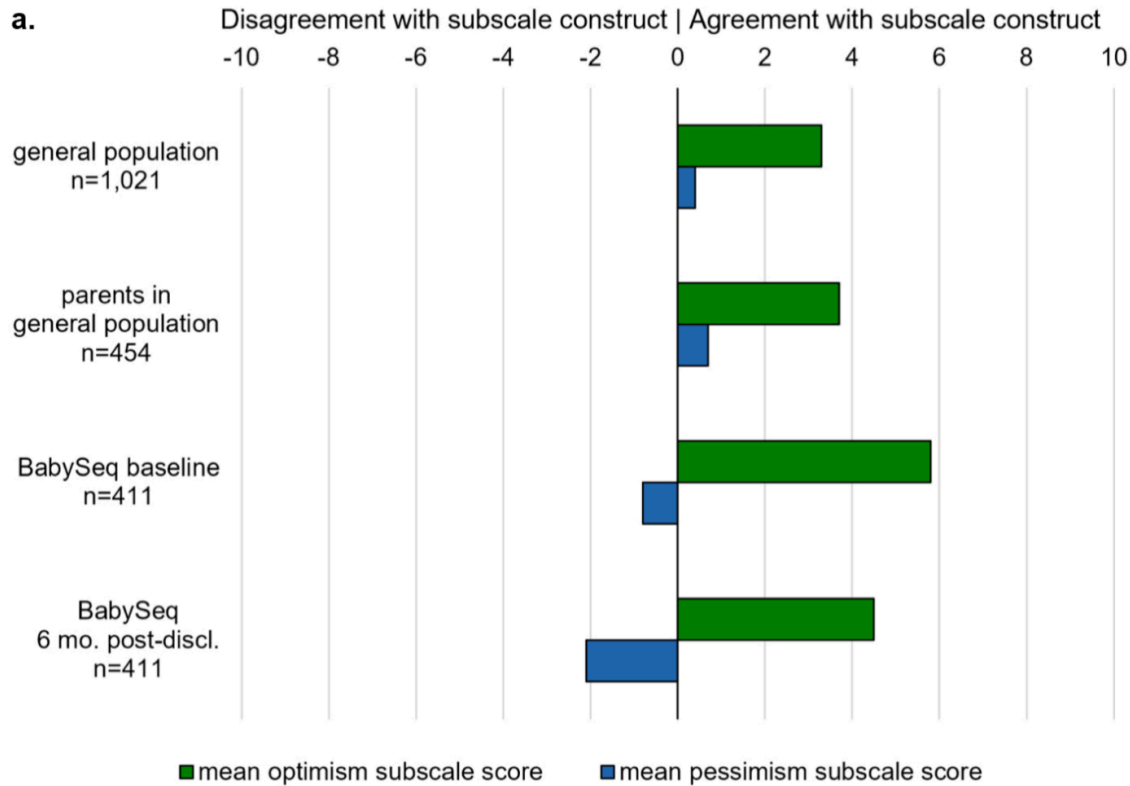


Figure 1

Genomic Orientation Scale

a. mean Genomic Orientation scale optimism and pessimism scores among the general population (n=1,021), parents within the general population (n=454), BabySeq parents at baseline (n=411), and BabySeq parents six months after results disclosure (n=411)

b. genomic orientation of BabySeq parents (n=411) at baseline and six months post-disclosure

Supplementary Files

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