

Psychosocial Factors Influencing Parental Interest in Genomic Sequencing of Newborns

Susan E. Waisbren, PhD,^{a,b} Caroline M. Weipert, MS, CGC,^c Rebecca C. Walsh, BS,^c Carter R. Petty, MA,^a Robert C. Green, MD, MPH^{b,c}

abstract

BACKGROUND: When parents of newborns are presented with the hypothetical possibility of obtaining genomic sequencing (GS) for their newborn infants immediately after birth, they express substantial interest. This study examined associations between expressed interest in GS and demographic and psychosocial variables some months after birth.

METHODS: A total of 1096 parents were enrolled in a study on GS of newborns shortly after the birth of their infants, before discharge from the postpartum floor. Of these parents, 663 (60.5%) completed a follow-up survey 2 to 28 months later that queried their interest in GS for their infant and whether they received worrisome health information during pregnancy, labor, and delivery. They were also administered the Parenting Stress Index. Multivariate logistic regression was used to examine factors associated with interest in GS of newborns.

RESULTS: Of parents, 76.1% indicated at least some interest in GS. A 10-point increase on the Parenting Stress Index was associated with an increase in the odds of having some interest in GS (odds ratio: 1.15; 95% confidence interval: 1.01–1.32). Age, gender, race, ethnicity, marital status, education, anxiety, and whether this was the first biological child were not significantly associated with interest in GS. Receiving worrisome health information was associated with greater interest in GS but this did not reach significance (odds ratio: 1.42; 95% confidence interval: 0.95–2.12).

CONCLUSIONS: This hypothetical survey study suggests that previous experiences leading to worrisome health information and parenting stress need to be considered when GS is offered. Additional research, currently underway, is exploring factors associated with real-life parental choices around whether to obtain GS of their newborns.



^aBoston Children's Hospital; ^bHarvard Medical School; and ^cDivision of Genetics, Department of Medicine, Brigham and Women's Hospital, Boston, Massachusetts

Dr Waisbren assisted in conceptualizing and designing the study, conducted data analyses, and drafted the initial manuscript; Dr Green conceptualized and designed the study, participated in data analyses, and reviewed and revised the manuscript; Ms Weipert participated in data entry and analyses, drafted portions of the initial manuscript, and reviewed and revised the manuscript; Ms Walsh conducted the interviews, participated in data entry and analyses, and reviewed and revised the manuscript; Mr Petty conducted the data analyses and reviewed and revised the manuscript; and all authors approved the final manuscript as submitted.

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Address correspondence to Susan E. Waisbren, PhD, Boston Children's Hospital, 300 Longwood Ave, Boston, MA 02115. E-mail: susan.waisbren@childrens.harvard.edu
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Clinical genomic sequencing (GS) is becoming more widespread as sequencing costs decrease and there are increasing indications for health-related genomic information.^{1,2} GS is frequently mentioned as a technology that may eventually be incorporated into the care of healthy individuals, perhaps someday as part of newborn screening.³ We previously reported that when parents of newborns were presented with the hypothetical possibility of GS for their newborns immediately after birth, 82% reported being at least somewhat interested.⁴ Here, we report an extension of this study in which parents of newborns were queried about their interest in GS for their infants some months after birth, and we examined the association between interest in GS and a number of demographic and psychosocial variables.

METHODS

Parents from our previous study on interest in GS immediately after the birth of a new infant were recontacted for this follow-up survey.⁴ These parents were originally enrolled within 48 hours of birth of a healthy newborn while at the well-baby newborn nursery at Brigham and Women's Hospital. A total of 663 of 1096 (60.5%) of these parents who enrolled in this study were surveyed 3 to 28 months after discharge from the well-baby nursery and asked on a 5-point scale if they were "not interested," "a little interested," "somewhat interested," "very interested," or "extremely interested" in GS for their infant. Parents were also asked a series of questions about their infant's health during pregnancy and the perinatal period as follows: "Were any of the following health problems present during the pregnancy or during labor and delivery? (please select all that apply): (a) problem found on ultrasound, (b) condition

suspected based on screening (blood test), (c) condition diagnosed by CVS [chorionic villus sampling], amniocentesis, or blood test, (d) problems with labor, (e) problems during delivery, (f) other health concerns or messages from OB [obstetric] doctor or nurse." If they selected any of those options the follow-up question was as follows: "Please provide more details on any answers you selected in this question."

Parental stress was measured by the self-report 36-item Parenting Stress Index (PSI)-Short Form, a well-validated measure that includes a Total PSI and 3 subscales: (1) Parental Distress, (2) Parent-Child Dysfunctional Interaction, and (3) Difficult Child. Higher scores indicate greater stress. As indicated in the test manual, scores in the 15th to 85th percentile constitute the "normal" range for the Total PSI. Parents who receive scores above the 90th percentile on the Total PSI are considered to be experiencing clinically significant parenting stress.^{5,6}

Two-sample *t* tests (2-tailed with α set at 0.05) were used to assess differences between mothers and fathers and between those who were rated as not interested versus interested in GS (defined as "not at all" or "only a little" interested versus "somewhat," "very," or "extremely" interested). Multivariate logistic regression was used to identify factors associated with interest in GS of newborns and included the following independent variables: age, gender, race (white versus other), ethnicity (Hispanic/Latino versus other) marital status (married versus other), education (some graduate school or higher), whether it was the parent's first biological child, child-related anxiety ("My baby makes me anxious"), having at least 1 health concern on the questions stated previously, and total stress (a

continuous measure in which 1 unit represents 10 PSI points).

The Partners Healthcare Human Research Committee approved the development and administration of this protocol. All participants provided informed consent.

RESULTS

Table 1 describes the sample and presents descriptive results from the survey. The sample included 445 mothers and 218 fathers. Among mothers, 85% were married, whereas 93% of fathers were married ($P < .01$). Participants were, on average, in their early 30s and in the upper middle class. They were generally well educated, with half reporting some graduate education or higher. Fifteen percent reported a family history of genetic disease. For 53% of fathers and 50% of mothers, this was their first biological child ($P < .05$). Compared with mothers, fathers were, on average, 2 years older ($P < .001$), and a lower percentage were identified as black/African American ($P < .05$).

Thirty-seven percent of mothers and 34% of fathers reported having received worrisome prenatal or perinatal information about their infant's health as defined above (see Table 1, Fig 1). For example, 73 parents (11.0%) reported that information from prenatal ultrasound examinations raised concerns, including such findings as "fluid on the kidney" or "intracardiac focus." Worrisome results from prenatal blood tests were reported by 13 parents (2.0%), most frequently tests suggesting a risk of Down syndrome. Amniocentesis/chorionic villus sampling-related health concerns were reported by 6 parents (1.0%) and included "cholestasis of pregnancy." Labor problems, noted by 53 parents (8.0%), were primarily reported as "failure to progress/dilate" or "cord around the neck." Problems with delivery were

reported by 70 parents (10.6%) and were most commonly noted as “meconium in the amniotic fluid” or “baby’s heart rate dropped.” Neonatal concerns reported by 20 parents (3.0%) included fever in their newborn, small size, and jaundice. Fifty-nine parents (8.9%) reported maternal health concerns that potentially placed the fetus at risk. Gestational diabetes was the most common of these health concerns.

Mean scores on the Total PSI were well within the normal range for the general population. For mothers and fathers, 8.9% and 9.2%, respectively, received a Total PSI score above the 85th percentile, and overall only 3.5% ($n = 23$) received scores above the 90th percentile. In terms of the PSI subscales, 14.3% of parents received scores above the 85th percentile on the Parental Distress subscale. Only 7.4% of parents received scores in this range on the Parent-Child Dysfunctional Interaction index and 7.1% of parents received scores above the 85th percentile on the Difficult Child subscale. With regard to anxiety, ~20% of mothers and fathers reported “agreeing” or “strongly agreeing” with the statement, “My baby makes me anxious.”

Mothers and fathers were equally interested in GS (Fig 2). Only 63 parents (9.5%) indicated that they had “no interest” and 13.1% expressed only “a little interest” in GS, whereas 33.5% of parents indicated that they were “somewhat interested,” 26.4% were “very interested,” and 16.2% were “extremely interested” in GS.

As noted in Table 2, *t* tests revealed no differences between parents expressing at least some interest in GS versus parents having little or no interest in GS on the basis of whether the parents received worrisome health information derived from prenatal ultrasound, blood tests, amniocentesis, or chorionic villus sampling. However, parents who

TABLE 1 Descriptive Characteristics of the Sample

Variable	Mothers ($n = 445$)	Fathers ($n = 218$)	Total Sample ($N = 663$)
Age, mean \pm SD (range), y	32.5 \pm 5.4 (15–50)	34.1 \pm 5.3*** (16–65)	33.1 \pm 5.4 (15–65)
Race, n (%)			
American Indian/Native	2 (0.4)	1 (0.5)	3 (0.4)
Asian	52 (12)	28 (13)	80 (12)
Black/African American	41 (9)*	8 (4)	49 (7)
Hawaiian/Pacific Islander	2 (0.4)	0 (0)	2 (0.3)
Other	66 (15)	28 (13)	94 (14)
White	291 (65)	159 (73)	450 (68)
Ethnicity, n (%)			
Hispanic or Latino	50 (11)	18 (8)	68 (10)
Hollingshead Index of Social Position ^a mean \pm SD (range)	3.7 \pm 1.5 (1–7)	3.9 \pm 1.2 (1–7)	3.7 \pm 1.4 (1–7)
Some graduate school or higher, n (%)	229 (51)	103 (47)	332 (50)
Married, n (%)	379 (85)	202 (93)**	581 (88)
Number of biological children, mean \pm SD (range)	1.7 \pm 0.8** (1–5)	1.5 \pm 0.7 (1–4)	1.6 \pm 0.8 (1–5)
First biological child, n (%)	221 (50)	130 (60)*	351 (53)
Family history of genetic disease, n (%)	35 (15)	21 (19)	56 (16)
PSI, ^b mean \pm SD			
Parental Distress	22.8 \pm 7.0	22.6 \pm 6.8	22.7 \pm 7.0
Parent-Child Dysfunctional Interaction	16.3 \pm 4.9	16.7 \pm 5.2	16.4 \pm 5.0
Difficult Child	20.1 \pm 6.0	20.6 \pm 6.4	20.3 \pm 6.1
Total PSI	59.2 \pm 15.3	59.8 \pm 16.2	59.4 \pm 15.6
“My baby makes me anxious” (≥ 2), n (%) ^c	85 (19)	51 (23)	136 (21)
Reporting at least 1 prenatal or perinatal health concern, n (%)	164 (37)	74 (34)	238 (36)
Somewhat, very, or extremely interested in GS, n (%)	338 (76)	174 (80)	512 (77)
Very or extremely interested in GS, n (%)	188 (42)	102 (47)	290 (44)

For comparisons between mothers and fathers: * $P \leq .05$, ** $P \leq .01$, *** $P \leq .001$.

^a Scale of 1–5, where low scores indicate higher social status.

^b Cutoff scores indicating “abnormal” levels of stress are as follows: Parental Distress, >30 ; Parent-Child Dysfunctional Interaction, >25 ; Difficult Child, >32 ; Total PSI, >85 .

^c >2 refers to responses indicating “Quite Often”, “Very Often” or “Always”

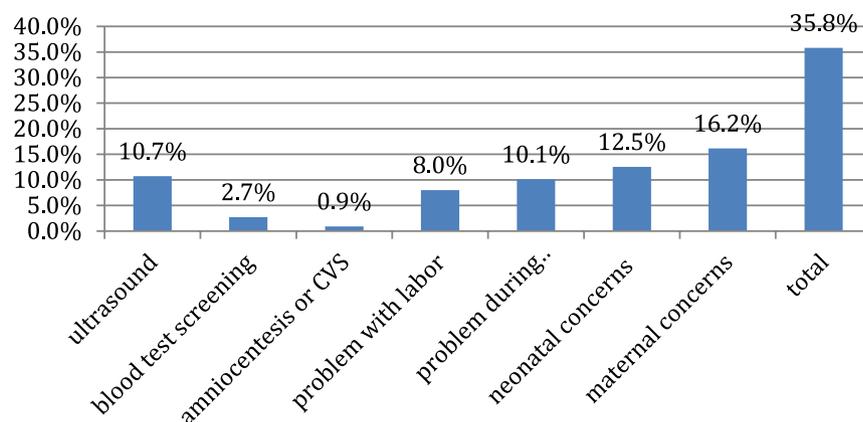


FIGURE 1

Concerns arising from conventional screening or clinical issues ($N = 662$). CVS, chorionic villus sampling. [medium]

reported problems during delivery or maternal health issues that potentially placed the child at risk were more likely to express interest in GS (defined as “somewhat,” very,” or “extremely” interested).

As shown in Table 3, logistic regressions examining factors associated with at least some interest in GS versus little or no interest in GS included the variables of age, gender, race, education, and whether this was their first biological child. None of these variables reached significance. Parents who indicated that their infant made them anxious were not more or less interested in GS. Total score on the PSI (examining each 10-point increase) was the only significant variable in the logistic regression (odds ratio: 1.15; 95% confidence interval: 1.01–1.32). Parents who reported at least 1 health concern had increased odds of being interested in GS, although this variable did not reach significance (odds ratio: 1.42; 95% confidence interval: 0.95–2.12).

DISCUSSION

This study examined factors associated with parents' reports of interest in GS for their newborn some months after they were discharged from the well-baby nursery. We found that 3 to 28 months after the birth of a newborn, 76% of parents were at least somewhat interested in GS. Demographic factors were not associated with level of interest in GS, and fathers and mothers responded similarly overall. Total PSI score was associated with greater interest in GS and there was a trend toward greater interest among parents who had experienced any worrisome health concerns in the prenatal or perinatal periods. The generalizability of our study is limited by the demographic characteristics of the population who agreed to the survey, because these parents were predominantly white, young, well-educated, and upper middle class.

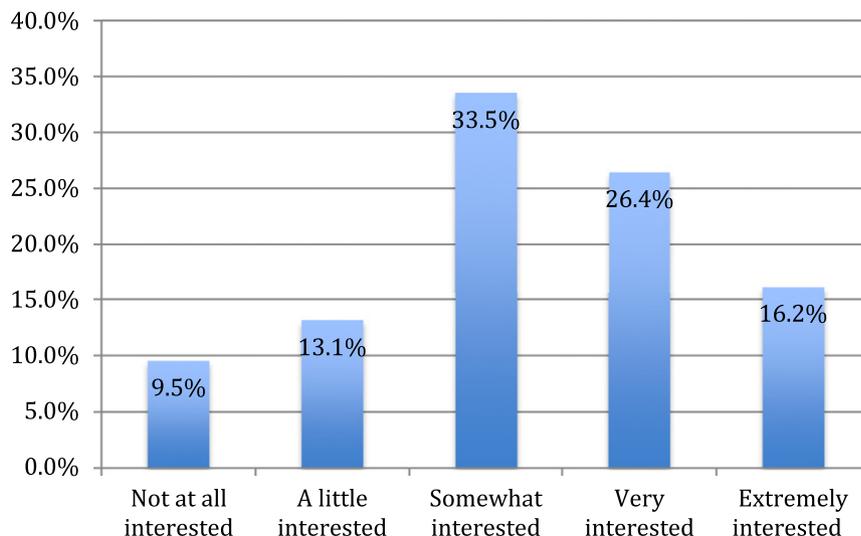


FIGURE 2
Percentage of parents interested in GS. [medium]

TABLE 2 PSI, Health Concerns, and Interest in GS

	Interested in GS ^a (n = 512)	Not Interested in GS ^b (n = 150)	P
PSI, mean raw score ± SD			
Parental Distress	23.0 ± 7.0	21.6 ± 6.8	.02
Parent-Child Dysfunctional Interaction	16.6 ± 4.9	15.9 ± 5.2	.16
Difficult Child	20.5 ± 5.9	19.5 ± 6.8	.08
Total PSI	60.1 ± 15.2	57.0 ± 16.5	.03
Health concerns, n (%)			
Problem noted on ultrasound	55 (10.7)	16 (10.7)	.98
Problem noted as result of blood test	11 (2.2)	1 (0.7)	.23
Problem noted as result of amniocentesis or chorionic villus sampling	6 (1.2)	0 (0)	.18
Problem during labor	44 (8.6)	9 (6.0)	.30
Problem during delivery	59 (11.5)	8 (5.3)	.03
Neonatal health concern	66 (12.9)	17 (11.3)	.61
Maternal problem putting infant at risk	91 (17.8)	16 (10.7)	.04
At least 1 health concern	193 (37.7)	44 (29.3)	.06

^a “Interested” was defined as “somewhat,” “very,” or “extremely” interested.

^b “Not interested” defined as “not at all” or “only a little” interested.

TABLE 3 Demographic Characteristics, Health Concerns, and Psychosocial Factors Associated With Parents Being at Least Somewhat Interested in GS for Their Infants

Variable	OR (95% CI)
Age	1.02 (0.98–1.07)
Female gender	0.80 (0.53–1.21)
White (versus other)	1.37 (0.89–2.10)
Hispanic or Latino (versus other)	1.58 (0.80–3.15)
Married	0.71 (0.36–1.37)
Some graduate school or higher	1.06 (0.71–1.58)
First biological child	1.17 (0.80–1.73)
“My baby makes me anxious”	1.22 (0.72–2.05)
Total PSI	1.15 (1.01–1.32)
Any health concerns	1.42 (0.95–2.12)

CI, confidence interval; OR, odds ratio.

Our results suggest that the high interest in GS among parents in the first 48 hours after birth that we previously documented⁴ is largely maintained among parents when they are home and during the first 2 years of their infants' lives. Our findings are also consistent with those of others. For example, Goldenberg et al⁷ surveyed a randomly selected sample of 1539 parents of children up to 18 years of age and reported high levels of hypothetical interest in GS for newborns, with 74% responding that they were at least somewhat interested. In a companion study, Dodson et al⁸ reported that parents whose youngest child had ≥ 2 health conditions were significantly more likely to be interested in GS for their child. Our findings support the idea that parents of newborns who report health concerns may be particularly interested in GS beyond the immediate postpartum period.

In our previous survey from parents in the immediate postnatal period, we reported an association between worrisome health information and less interest in GS for their newborn infants.⁴ Our survey of parents 3 to 28 months after birth suggests a trend in the opposite direction. Our data do not allow us to comment

on the specific motivations of the parents, but screening actions are generally taken to prevent adverse health outcomes or seek reassurance. Individuals seeking reassurance but who perceive themselves to be at risk tend to reject screening.⁹ In our case, parents who had received worrisome health information in the prenatal/perinatal period but whose infants had subsequently developed in good health might no longer be concerned about discovering a negative outcome, and therefore may have been more interested in GS.

With the steady increase in prenatal screening of all types, parents will continue to receive concerning information about their unborn infant.^{10,11} Stress levels may also increase in the neonatal period due to the expansion of conventional newborn screening, with its concomitant increase in false-positive identifications and findings of uncertain significance.^{12–15} Our results suggest that parental stress and worrisome prenatal/perinatal experiences will influence parental interest in GS when it is offered. Additional research on factors motivating parents to accept or reject GS will clarify how and when parents should be approached.

Although these data were generated from a hypothetical survey, we are now engaged in assessing whether interest in GS will be similar when parents of newborns are presented with the real option of having their infant's genome sequenced. The ongoing National Institutes of Health-funded BabySeq Project (<http://www.genomes2people.org/babyseqproject>) is exploring the interest and outcomes of sequencing newborns. Information derived from these studies will influence newborn screening policies and programs that may include GS in the future.

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ABBREVIATIONS

GS: genomic sequencing
PSI: Parenting Stress Index

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