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# In the Early Postpartum Period, Parents are Interested in Newborn Genomic Testing

**Susan E. Waisbren, PhD**[1,2,3], **Danielle K. Bäck, BS**[3,4], **Christina Liu, BS**[4], **Sarah S. Kalia, ScM, CGC**[4], **Steven A. Ringer, MD, PhD**[3,5], **Ingrid A. Holm, MD, MPH**[1,3,6], and **Robert C. Green, MD, MPH**[3,4,7]

[1]Division of Genetics and Genomics, Boston Children's Hospital, Boston, MA

[2]Department of Psychiatry, Boston Children's Hospital, Boston, MA

[3]Harvard Medical School, Boston, MA

[4]Division of Genetics, Brigham and Women's Hospital, Boston, MA

[5]Department of Newborn Medicine, Brigham and Women's Hospital, Boston, MA

[6]Manton Center for Orphan Diseases Research, Boston Children's Hospital, Boston, MA

[7]Partners Personalized Medicine, Boston, MA

## Abstract

**Purpose—**We surveyed parents to ascertain interest in newborn genomic testing and determine whether these queries would provoke refusal of conventional newborn screening (NBS).

**Methods—**After brief genetics orientation, parents rated their interest in receiving genomic testing for their healthy newborn on a 5-point Likert scale and answered questions about demographics and health history. We used logistic regression to explore factors associated with interest in genomic testing and tracked any subsequent rejection of NBS.

**Results—**We queried 514 parents within 48 hours after birth while still in the hospital (mean age (sd) 32.7 (6.4) years, 65.2% female, 61.2% white, 79.3% married). Parents reported being not at all (6.4%), a little (10.9%), somewhat (36.6%), very (28.0%) or extremely (18.1%) interested in genomic testing for their newborns. None refused conventional NBS. Married participants and those with health concerns about their infant were less interested in newborn genomic testing (p=0.012 and p=0.030, respectively). Mothers' and fathers' degree of interest was discordant (≥ 2 categories different) in 24.4% of couples.



Corresponding author: Robert C. Green, MD, MPH, Brigham and Women's Hospital and Harvard Medical School, Partners Center for Personalized Genetic Medicine, EC Alumnae Building, Suite 301, 41 Avenue Louis Pasteur, Boston, MA 02115, (office) 617-264-5834, (fax) 617-264-3018, (cell) 617-966-3216, ; Email: rcgreen@genetics.med.harvard.edu

**DISCLOSURES**

The authors declare no conflict of interest.

**Conclusions—**Interest in newborn genomic testing was high among parents of healthy newborns and the majority of couples had similar levels of interest. Surveying parents about genomic sequencing did not prompt rejection of NBS.

### Keywords

newborn screening; newborn genomic testing; sequencing

## INTRODUCTION

Next generation whole exome and genome sequencing is currently being integrated into clinical diagnostics,[1] and there is active debate about the degree to which sequencing may be useful for screening or predispositional testing in adults and in children.[2] In particular, researchers have begun exploring how sequencing could be utilized to provide personalized health information in the newborn period.[3–6] As the potential for screening newborns with genomic testing would be of greatest value if provided shortly after birth, it is important to study parents' attitudes towards such testing immediately post-partum. We therefore explored parental interest in newborn genomic testing in the well newborn nursery prior to discharge from the hospital. We assessed predictors of parental interest and analyzed concordance between parental preferences for this testing.

We also evaluated the impact of asking these question on possible rejection of state-mandated newborn screening (NBS). NBS is one of the most established and successful public health programs in the world. Each year, thousands of infants who would develop devastating or life threatening conditions are identified and treated before symptoms occur.[5,7] Approximately 98% of parents of the 4.3 million infants born each year in the United States participate in NBS, which in most states is administered without formal consent, but with some provisions for opt-out.[8] Despite the high participation rate, most women with children ages 10 or younger do not recall receiving information or being aware that they had any choice about NBS.[9] The high proportion of unawareness about NBS, in combination with the opt-out consent model utilized in most states, has raised concerns that any additional options, procedures or even discussions about genomics in the immediate post-partum period could create confusion and prompt rejection of NBS. To address this concern, we also monitored the parents who responded to our questions during the remainder of their stay in hospital to determine if we could observe any association between our questions and their participation in NBS.

## MATERIALS AND METHODS

### Study Design

Between July, 2012 and December, 2013 and with permission of the charge nurse, research assistants (RA) approached parents in the well baby nursery at Brigham and Women's Hospital within 48 hours following the birth of a healthy newborn. Individuals who did not speak English, had impaired-decision making capacity, or had a newborn in the NICU were excluded. The RA explained that our survey was examining parental attitudes toward a "test that is not yet being done for healthy babies," that was different from the state-mandated

heel stick blood test. Those who declined to participate in the study were asked to provide their gender, age, and the highest completed level of education. Of the 1096 parents who consented to participate in the study, 582 were randomly selected for an intervention at a later time point and will be described in a subsequent publication.

The remaining 514 parents received a brief introduction to the genome, inheritance patterns, genetic risk, and implications for health and clinical care. After answering demographics questions, parents were asked to imagine that they were offered "a chance to take part in a research study that would test many or all of the genes in their baby" where they would receive the results, and rate their interest in this newborn test on a 5-point Likert scale. Parents also responded "yes" or "no" to the following questions: "Are there any health concerns with your baby?" and "Has a doctor diagnosed anyone in your family with a genetic disease?"

When both parents were surveyed, no attempt was made to separate the parents, so they were able to hear each other's responses. Concordance between parents who were part of a couple was defined as both parents reporting a similar level of interest in newborn genomic testing, i.e. within 1 unit on the 5-point Likert scale; discordance was defined as parents' answers differing by 2 units or more.

In order to determine if any parents refused NBS, we took advantage of the fact that the nursury staff utilizes an established system for tracking NBS at Brigham and Women's Hospital, ensuring that all newborn blood samples are received by the state laboratory that performs NBS. As part of this system, all missing samples are investigated and any refusals are recorded. The Partners Healthcare institutional review board approved the development and administration of this protocol. The study was registered with clinicaltrials.gov (NCT01736501).

### Statistical Analysis

Logistic regression was used to explore associations of parental interest in newborn genomic testing with demographics, family history of genetic disease, and parental health concerns about their baby. Within each couple, a concordance analysis was also performed by comparing the parents' level of interest in newborn genomic testing, as reported on the 5-point Likert scale, and identifying the percentage of couples who were concordant or discordant. Logistic regression was applied to identify associations between concordance and demographics, family history of genetic disease, and newborn health concerns. Data were analyzed using Stata 13.1 (StataCorps, College Station, TX).

## RESULTS

Of 1309 parents approached, 1096 parents (83.7%) agreed to participate in the study, and 514 were randomized to receive the survey in hospital as reported here. Table 1 describes the characteristics of the 514 parents who consented and completed the survey questions. Table 1 also displays the results of the logistic regression analysis examining the effect of each descriptive feature on parental interest in newborn genomic testing as part of a research study. Participants who reported health concerns in their infant (OR: 0.39, 95% CI: 0.16–

0.91, p=0.030) or were married (OR: 0.36, 95% CI: 0.16–0.80, p=0.012) were less likely to express interest in newborn genomic testing. Parental interest in newborn genomic testing was not significantly associated with age, gender, race, ethnicity, level of education, being a first-time biological parent, or family history of genetic disease.

Figure 1 depicts the percentages of parents expressing their level of interest on a 5-point Likert scale. The majority (82.7%) of parents reported being somewhat, very, or extremely interested in newborn genomic testing.

Among couples, a concordance analysis was performed to determine if mothers and fathers in the same family unit reported similar attitudes towards genomic newborn testing in a research setting. Of the 168 couples in which both parents were surveyed, 127 couples (75.6%) were concordant in their responses, whereas 41 couples (24.4%) were discordant. Out of the 41 couples that were discordant, the male respondents were more interested in newborn genomic testing in 23 couples. Concordance was more likely if the couple was married (OR: 2.85, p=0.012).

Over the two-year study period, none of the parents surveyed about genomic newborn screening refused routine state-mandated NBS.

## DISCUSSION

Our study shows that the majority of parents on the newborn unit were interested in hypothetical genomic testing of their newborns within a research study. As detailed above, respondents who reported newborn health concerns at the time of the survey were less likely to express interest in newborn genomic testing. It is possible that parents who reported that their child had a health concern were more stressed and less interested in pursuing genetic testing due to its potential to increase their emotional distress. Additionally, parents who had faced what they perceived to be a health problem in their newborn may not have wished to discover a hereditary component to that problem, or may have felt sufficiently stressed by this that they felt unwilling to take on new information. Parental interest among unmarried couples was elevated in comparison to married couples, perhaps because unmarried couples are less traditional, even though interest in newborn genomic testing was high among overall survey participants.

Of interest, gender, age, race, ethnicity, level of education, family history of genetic disease, and whether or not the infant was a first-born child were not significantly associated with levels of parental interest in newborn genomic testing. These data suggest that if newborn genomic testing becomes available, there would be robust interest among parents of newborns, regardless of demographics.

Among participants where both parents were surveyed, most parents reported similar levels of interest in newborn genomic testing. This suggests that if newborn genomic testing were offered in a research setting, parents would be likely to agree, although strategies need to be in place to ensure that both parents provide informed consent.

Our survey has several limitations. First, this study relied on participant responses to a hypothetical opportunity to receive newborn genomic testing as part of a research study – actual testing was not offered. Additionally, we did not restrict communication of mothers and fathers who were participating in the study together and this may have artificially enhanced concordance. Study strengths include a large size and diverse ethnicity of participants.

The results from sampling this relatively diverse cohort align closely with recent research indicating that there is broad public support for genetic testing of infants. One study found that parents strongly supported newborn genetic testing, with 69% believing that testing should be available for any condition.[10] In another study, 74% of parents with children under the age of 18 were either definitely or somewhat interested in whole-genome sequencing as part of a NBS program.[11] Thus, parental decisions relatively soon after birth are congruent with these findings, despite fatigue and other stressors they may be experiencing.[12]

Some clinicians, public health experts, ethicists, and legislators have expressed concern that newborn genomic testing - and the consent process that would accompany it – could confuse parents and undermine the established NBS program.[4,13] There is concern that even raising the possibility of sequencing technologies in a research context could cause parents to refuse to state-mandated newborn screening, inadvertently risking harm to their infants.[3] One recent study of Canadian residents found that a lower proportion of parents (80%) reported willingness to participate in screening using genomic technologies as compared with screening using current technologies (94%), perhaps because of concerns about genetic privacy.[14] Preliminary data from research conducted in Scotland found that requiring formal informed consent for a new cystic fibrosis test resulted in a 0.033% to 0.072% increase in rejection of all screening tests.[15] However, in our study, none of the 514 parents who were asked about their interest in newborn genomic testing challenged or rejected state-mandated newborn screening for their babies. While we only inquired about interest in genomic testing and did not actually offer this new technology, our findings suggest that discussing newborn genomic testing with parents shortly after birth does not provoke confusion or refusals of state-mandated newborn screening.

Additional concerns with genetic newborn testing within the scientific community focus on public health issues. Technological advances in whole genome sequencing could significantly increase the number of infants identified with potential disorders, and the health system might not be prepared to provide adequate follow-up.[7] Increases in false positive or inconclusive findings could also lead to increased parental stress or dysfunctional parent-child relationships[4,16–19] and the expenditure of additional health-care costs.[20] Future studies should examine discordance in parental attitudes in newborn genomic testing, effective counseling strategies for couples interested in genomic testing, and the psychological, medical and health utilization consequences of receiving actual newborn genomic testing results. With the cost of genome sequencing continuing to fall rapidly, its utilization in the newborn period may increase relatively soon. Our results emphasize considerable interest of parents in hospital for obtaining genomic testing for their newborns.

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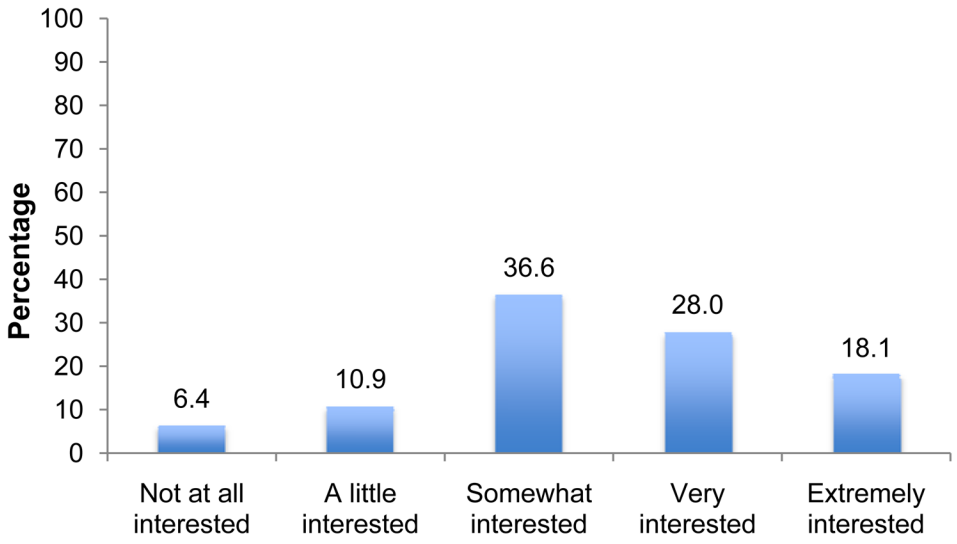


**Figure 1.**
Parental interest, immediately after birth, in hypothetical newborn genomic testing for their newborns as part of a research study.

## Table 1

Participant demographics and results of logistic regression assessing the association of parental interest in newborn genomic testing as part of a research study, controlling for all variables listed in the Table.

| Variable | In-Patient Cohort (n=514) | OR (95% CI) | p |
|---|---|---|---|
| Mean age ± sd (range) | 32.7 ± 6.4 (15–65) | 1.05 (1.00 – 1.10) | 0.066 |
| Female, n (%) | 335 (65.2) | 1.03 (0.61–1.72) | 0.917 |
| White, n (%) | 314 (61.2) | 1.53 (0.89 – 2.62) | 0.123 |
| Hispanic or Latino, n (%) | 64 (12.5) | 0.94 (0.43 – 2.05) | 0.882 |
| Married, n (%) | 407 (79.3) | 0.36 (0.16 – 0.80) | 0.012 |
| Some graduate school or higher, n (%) | 248 (48.3) | 0.87 (0.51 – 1.48) | 0.611 |
| First biological child, n (%) | 270 (52.7) | 1.44 (0.89 – 2.33) | 0.142 |
| Family history of genetic disease, n (%) | 70 (13.7) | 0.85 (0.42 – 1.73) | 0.655 |
| Infant health concerns, n (%) | 29 (5.7) | 0.39 (0.16 – 0.91) | 0.030 |