BRIEF REPORT | Genetics inMedicine

# Parents are interested in newborn genomic testing during the early postpartum period

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**Purpose:** We surveyed parents to ascertain interest in newborn genomic testing and determine whether these queries would provoke refusal of conventional state-mandated newborn screening.

**Methods:** After a 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 newborn screening.

**Results:** We queried 514 parents within 48 hours after birth while still in 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 state-mandated newborn screening. 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). Degree of interest for mothers and fathers was discordant (at least two categories different) for 24.4% of couples.

**Conclusion:** 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 newborn screening.

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**Key Words:** newborn genomic testing; newborn screening; sequencing

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 children or adults.[2] In particular, researchers have begun exploring how sequencing could be utilized to provide personalized health information in the newborn period.[3–6] Because 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 toward such testing immediately postpartum. We therefore explored parental interest in newborn genomic testing in the well newborn nursery before 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 questions 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 newborns 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 newborns 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 aged 10 years or younger do not recall receiving information or being aware that they had any choice about NBS.[9] This degree of unawareness about NBS, in combination with the opt-out consent model utilized in most states, has raised concerns that discussions about genomics in the immediate postpartum period could create confusion and prompt rejection of NBS. To address this concern, we also monitored the parents who participated in our study during the remainder of their stay in hospital to assess any association between our questions and their participation in NBS.

## MATERIALS AND METHODS

### Study design

Between July 2012 and December 2013, research assistants approached parents in the well baby nursery at Brigham and Women's Hospital within 48 hours after the birth of a healthy newborn. Individuals who did not speak English, had impaired decision-making capacity, or had a newborn in the neonatal intensive care unit were excluded. The research assistants explained that our survey examined 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.

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Of the 1,096 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," told that they would receive the results, and asked to 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, an attempt was made to separate the parents, so they would not 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.

To determine whether any parents refused NBS, we used the established system for tracking NBS at Brigham and Women's Hospital, which ensures 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. For each couple, a concordance analysis was also performed by comparing the parents' levels of interest in newborn genomic testing, as reported on the 5-point Likert scale, and by 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 1,309 parents approached, 1,096 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 for their newborn (odds ratio: 0.39, 95% confidence interval: 0.16–0.91, $P = 0.030$) or who were married (odds ratio: 0.36, 95% confidence interval: 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.

**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* value |
|---|---|---|---|
| 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 |

CI, confidence interval; OR, odds ratio.

**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 whether mothers and fathers in the same family unit reported similar attitudes toward 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. Of the 41 couples who were discordant, the male respondents were more interested in newborn genomic testing in 23 couples. Concordance was more likely if the couple was married (odds ratio: 2.85, $P = 0.012$).

Over the 2-year study period, none of the parents surveyed about genomic NBS 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. 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 genetic testing because of 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 illness, or may have felt sufficiently stressed by this that they felt unwilling to take on new information. Parental interest

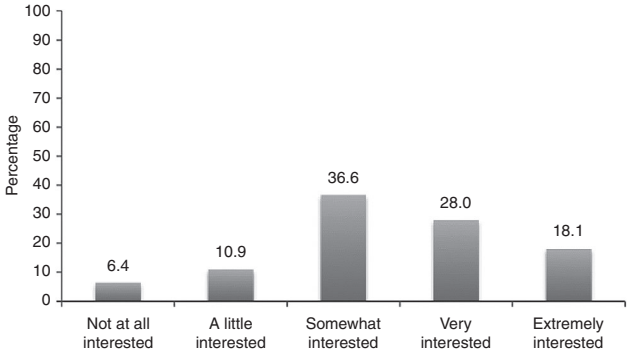


**Figure 1 Parental interest, immediately after birth, in hypothetical newborn genomic testing for their newborns as part of a research study.**

among unmarried couples was elevated in comparison with that among married couples, perhaps because unmarried couples are less traditional.

Of interest, gender, age, race, ethnicity, level of education, family history of genetic disease, and whether the newborn 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, then there would be robust interest among parents of newborns, regardless of demographics.

When 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, then 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 fully restrict communication of mothers and fathers who were participating in the study together, and this may have artificially enhanced concordance. Study strengths include 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 newborns. 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 younger than age 18 years 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 NBS, inadvertently risking harm to their newborns.[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 appears to have increased parental rejection of all screening tests from a level of 0.033 to 0.072%. The study found an additional 0.002% increase in rejection of one or more of the individual screening tests following implementation of informed consent for cystic fibrosis testing.[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 NBS for their babies. Although 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 soon after birth does not provoke confusion or refusal of state-mandated NBS.

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 newborns 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 toward newborn genomic testing, effective counseling strategies for couples interested in genomic testing, and the psychological, medical, and health-utilization consequences of receiving newborn genomic testing results. With the cost of genome sequencing continuing to decrease rapidly, its utilization during the newborn period may increase relatively soon. Our results emphasize considerable interest of parents in the hospital in obtaining genomic testing for their newborns.

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## DISCLOSURE

The authors declare no conflict of interest.

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