Parents Are Interested in Newborn Genomic Testing in the Early Postpartum Period

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Scholarly report submitted in partial fulfillment of the MD Degree at Harvard Medical School

March 30, 2017
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Parents are Interested in Newborn Genomic Testing in the Early Postpartum Period

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Abstract

Purpose: We surveyed new parents at Brigham and Women’s Hospital in the early postpartum period to assess their interest in newborn genomic testing. We also assessed whether these survey questions affected uptake of state-mandated newborn screening.

Methods: After providing consent, parents received a brief orientation to genetics. They then 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. We also evaluated concordance between couples’ survey responses.

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.

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 newborn screening.
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Glossary:
WES: Whole Exome Sequencing
WGS: Whole Genome Sequencing
NBS: Newborn Screening
Section 1: Introduction

Whole exome sequencing (WES) and whole genome sequencing (WGS) are becoming more integrated into clinical care, as physicians realize the potential that these diagnostics have to diagnose rare genetic diseases. Researchers have already examined how WES and WGS might improve clinical care for adult patients, but are now assessing how WES and WGS may be integrated into newborn care in the early postpartum period. In order to understand the feasibility of implementing newborn genomic screening, we surveyed new parents in the early postpartum period to evaluate their attitudes towards and interest in this genetic testing. We also tracked refusals in state-mandated newborn screening (NBS) and assessed concordance between couples’ survey responses.
Section 2: Student Role

As a summer research intern, I had several roles on this research project. First, I performed a literature review on whole exome sequencing and whole genome sequencing for newborns, which facilitated the preparation of our manuscript. Second, I performed data management and analysis as outlined in the methods section, using STATA 13.1. Third, along with another researcher, I drafted the abstract and manuscript, and incorporated edits from the rest of the research team. Finally, in order to ensure that the response rate for the 582 parents who were randomized to a later survey intervention was adequate, I called and surveyed participants who had not yet completed their follow up surveys.
Section 3: Methods

Between July 2012 and December 2013, research associates consented 1,096 parents in the well baby nursery at Brigham and Women’s Hospital within 48 hours of the birth of a healthy child. 582 were randomized for a later intervention, which will be analyzed for a subsequent project. The other 514 parents received a brief orientation to genetics, including genetic risk, inheritance patterns, and health implications of genetic testing. They then 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. We also evaluated concordance between couples’ survey responses, which was defined as responding within 1-point on the 5-point Likert scale. Data were analyzed using Stata 13.1 The Partners Healthcare Institutional Review Board approved the development and administration of this protocol. The study was registered with clinicaltrials.gov (NCT01736501).
Section 4: Results

We surveyed 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). Overall, 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 (Figure 1). Demographics for survey participants and results for the logistic regression to evaluate factors associated with parental interest in newborn genomic testing are reported in Table 1. 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. During the two year study, no participants refused state-mandated newborn screening.
Section 5: Discussion, Limitations, Conclusions and Suggestions for Future Work

These results show 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.

Interestingly, 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 had 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.

Future research should examine the impact of newborn genomic screening on health systems, which may not be adequately prepared for the potentially increased number of newborns identified with genetic disorders. Additionally, future studies should examine counseling strategies for couples considering newborn genomic testing, as well as the effects of receiving newborn genomic testing on healthcare utilization.
Section 6: Acknowledgments

I would like to acknowledge the entire BabySeq Project team for their support and encouragement throughout the project!
References


### Tables and Figures

**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.

<table>
<thead>
<tr>
<th>Variable</th>
<th>In-Patient Cohort (n=514)</th>
<th>OR (95% CI)</th>
<th>p</th>
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<tbody>
<tr>
<td>Mean age ± sd (range)</td>
<td>32.7 ± 6.4 (15-65)</td>
<td>1.05 (1.0 – 1.10)</td>
<td>0.066</td>
</tr>
<tr>
<td>Female, n (%)</td>
<td>335 (65.2)</td>
<td>1.03 (0.61-1.72)</td>
<td>0.917</td>
</tr>
<tr>
<td>White, n (%)</td>
<td>314 (61.2)</td>
<td>1.53 (0.89 – 2.62)</td>
<td>0.123</td>
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<tr>
<td>Hispanic or Latino, n (%)</td>
<td>64 (12.5)</td>
<td>0.94 (0.43 – 2.05)</td>
<td>0.882</td>
</tr>
<tr>
<td>Married, n (%)</td>
<td>407 (79.3)</td>
<td>0.36 (0.16 – 0.80)</td>
<td>0.012</td>
</tr>
<tr>
<td>Some graduate school or higher, n (%)</td>
<td>248 (48.3)</td>
<td>0.87 (0.51 – 1.48)</td>
<td>0.611</td>
</tr>
<tr>
<td>First biological child, n (%)</td>
<td>270 (52.7)</td>
<td>1.44 (0.89 – 2.33)</td>
<td>0.142</td>
</tr>
<tr>
<td>Family history of genetic disease, n (%)</td>
<td>70 (13.7)</td>
<td>0.85 (0.42 – 1.73)</td>
<td>0.655</td>
</tr>
<tr>
<td>Infant health concerns, n (%)</td>
<td>29 (5.7)</td>
<td>0.39 (0.16 – 0.91)</td>
<td>0.030</td>
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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.