

# The BabySeq Project: Genome Sequencing for Childhood Risk and Newborn Illness

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# Pilot Survey Protocol

(N = 1309)

Approach parents within 72 hours of delivery  
Consent parent(s) and randomize family unit to baseline survey

(N = 582)

- Demographics
- Consent to re-contact

(N = 514)

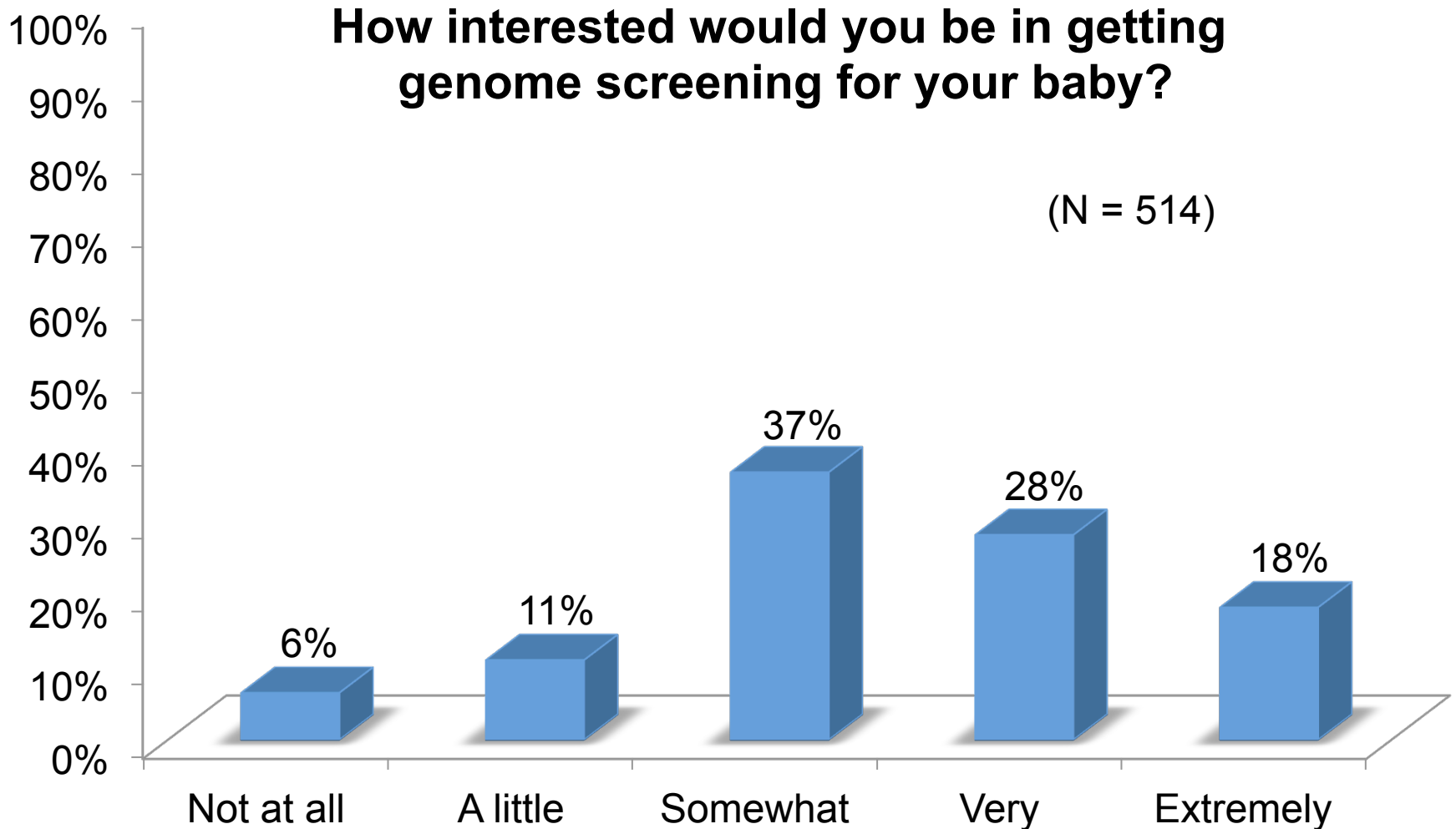
- Demographics
- Consent to re-contact
- Genetics orientation
- Interest in genome screening

3-24 months

(N > 605)

Genetics orientation and randomize to follow-up survey  
with or without mock genomic results

# Parental Interest in the Hospital



# Demographics and Associations with Parental Interest at Baseline

Variable	In-Patient Cohort (n=514)	OR (95% CI)	p
Mean age $\pm$ sd (range)	32.7 $\pm$ 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
<b>Married, n (%)</b>	<b>407 (79.3)</b>	<b>0.36 (0.16 – 0.80)</b>	<b>0.012</b>
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
<b>Infant health concerns, n (%)</b>	<b>29 (5.7)</b>	<b>0.39 (0.16 – 0.91)</b>	<b>0.030</b>

# Concordance Analysis

- 168 couples (among 514 parents) rated their interest in newborn genome screening
- 127 couples (76%) reported similar levels of interest
- 41 couples (24%) were discordant in their views
  - Concordance more likely if the couple was married (OR: 2.85,  $p=0.012$ )

# Limitations

- Participants rated their hypothetical interest. Actual uptake was not measured.
- Mothers and fathers queried around the same time, perhaps influencing responses
- Increased anxiety or confusion about NBS was not tracked

# The BabySeq Project

(U19 HD077671)

- First randomized trial to explore benefits and risks of genome sequencing (GS) in healthy and sick newborns
- 240 healthy newborns
- 240 NICU newborns
- Parents and physicians
- Timeline: 2013-2018



# BabySeq Project Overview

Pre-Enrollment Genetic Counseling,  
Consent, Blood Draw, Family History with Genetic Counselor

240 Healthy Newborns at BWH and Parents

- Standard of Care NBS
- Family History

- Standard of Care NBS
- Family History
- Genome Report

240 Newborns in NICU at BCH and Parents

- Standard of Care NBS
- Family History

- Standard of Care NBS
- Family History
- Genome Report

*Optional:*  
•Indication-Based Report

Consultation and Results Disclosure with Genetic Counselor and Study Physician.  
Consultation Note and Testing Reports placed in Medical Record  
and sent to other care providers

10-month Follow-up Appointment and Exam with Study Physician and  
Genetic Counselor

Medical Record Review

Outcomes collected. Study Physicians and GCs available for  
questions from parents, NICU MDs and outside MDs



# Workflow for Infant and Parents



Study Physicians and Genetic  
Counselors available for Qs

# Workflow for Physicians

Neonatologists, NICU Specialists, Community Pediatricians receive  
Baseline/Attitudes Survey (completion = consent)

None of MD's patients  
enroll

MD's patient(s) enroll,  
randomized to  
NBS-only

MD's patient(s) enroll,  
at least 1 randomized  
to GS+NBS

Baseline Survey reminder if incomplete

Receive summary of results disclosure  
by study GC to family.  
Option to discuss with study GC/MD

Post-Disclosure/  
Utilization Survey  
(completion = consent)

End of Study/Attitudes Survey  
(completion = consent)

Study MDs and GCs available for questions

# Data Collection Domains

	Parents MDs		Parents		MDs
Survey Domain	Baseline	Post-Disclosure	3 month Follow-up	10 month Follow-up	End of Study
Attitudes	X X		X		X
Perceived Utility	X X	X X	X	X	X
Healthcare Utilization	X X	X X		X	X
Health Behaviors & Intentions	X	X	X	X	
Parent-Child Relationship	X	X	X	X	
Perceptions of Child	X		X	X	
Personal Distress	X	X	X	X	
Partner Relationship	X		X	X	

# The BabySeq Project Team

## Leadership

Alan H. Beggs, PhD (Joint PI)  
Robert C. Green, MD, MPH (Joint PI)  
Peter J. Park, PhD  
Heidi L. Rehm, PhD  
Tim W. Yu, MD, PhD  
Pankaj B. Agrawal, MD, MMSC  
Richard B. Parad, MD, MPH  
Ingrid A. Holm, MD, MPH  
Amy L. McGuire, JD, PhD

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# Thank You

Questions?

Comments?



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