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

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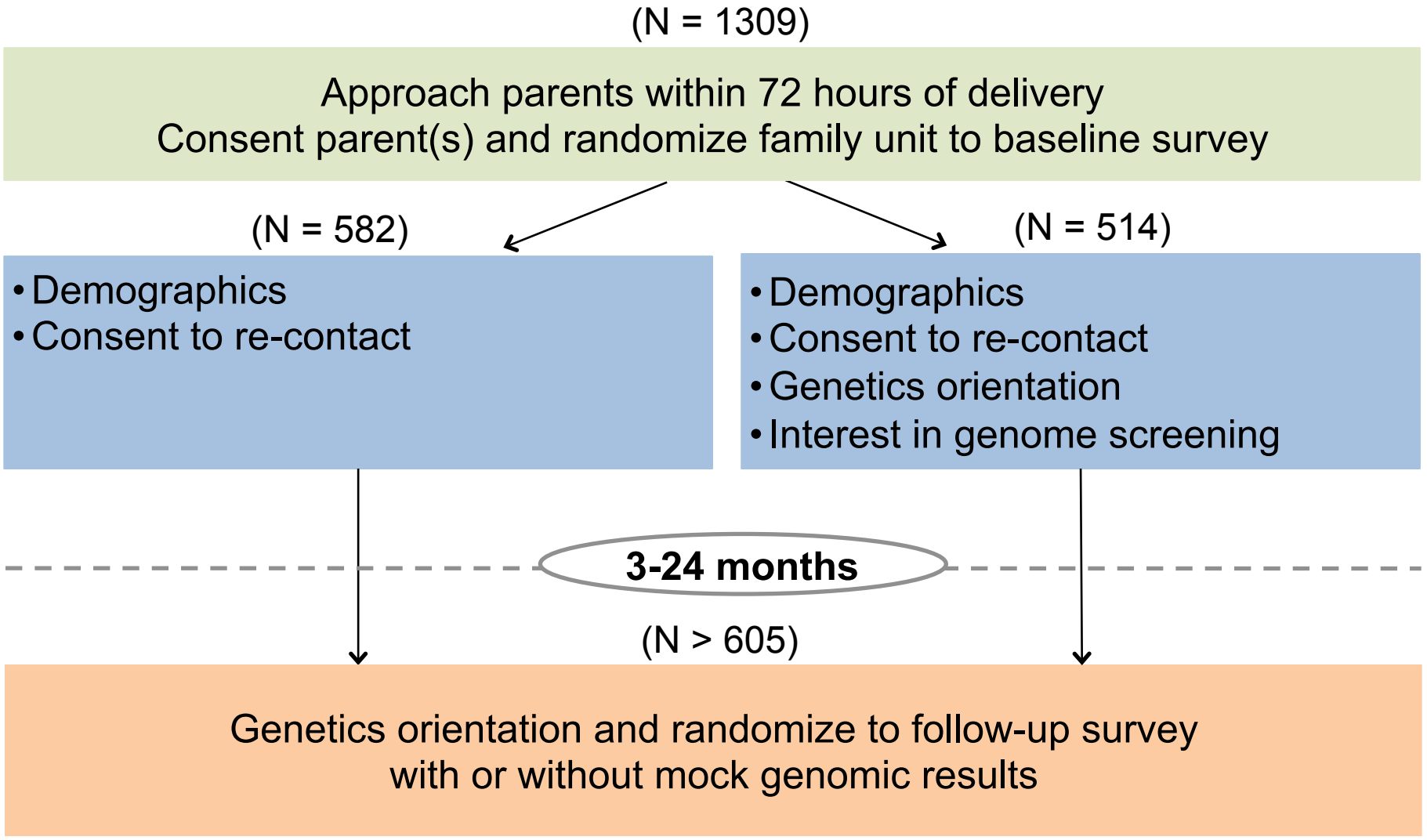
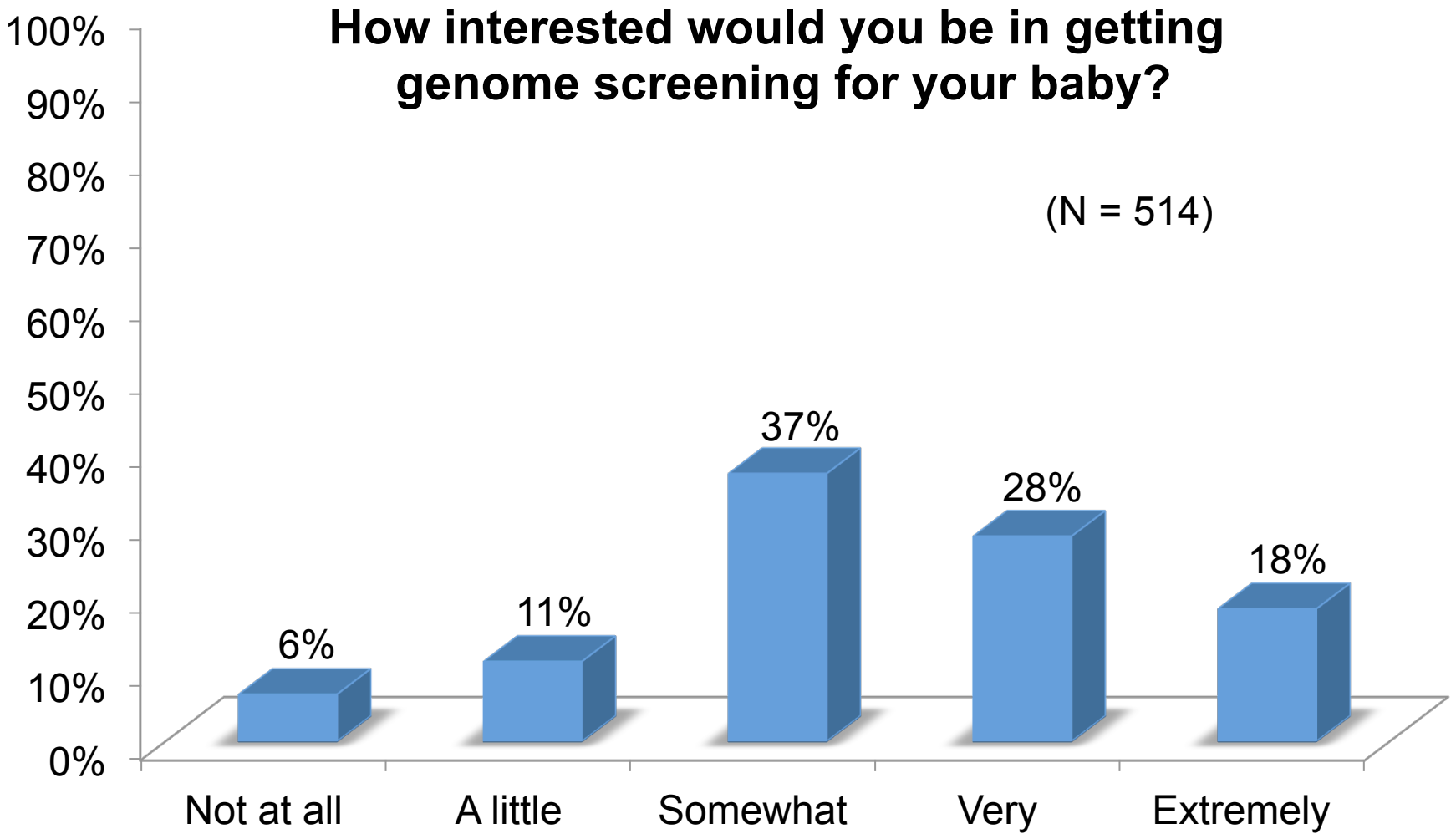
Pilot Survey Protocol



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 ± 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

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)

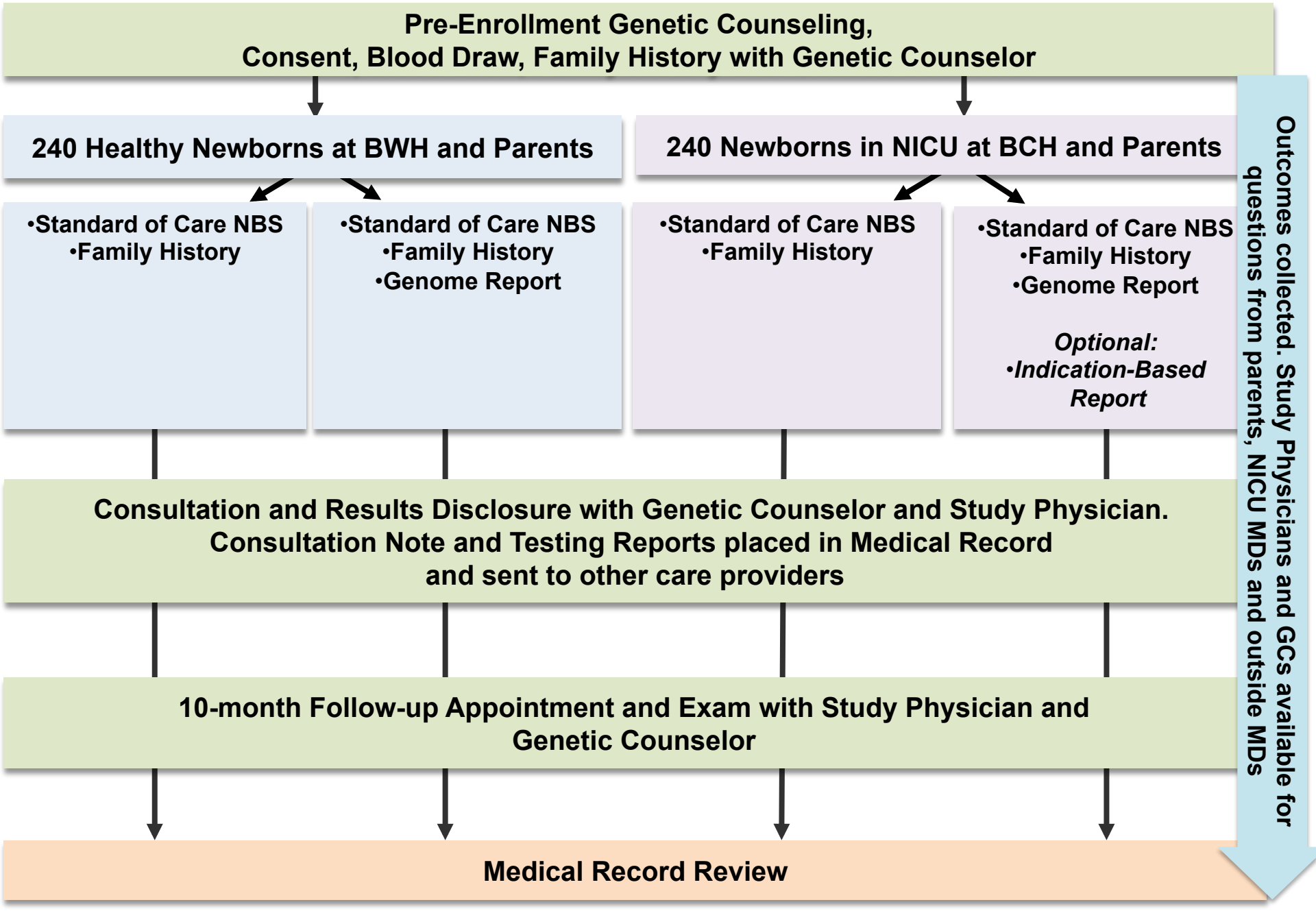
Limitations

- 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

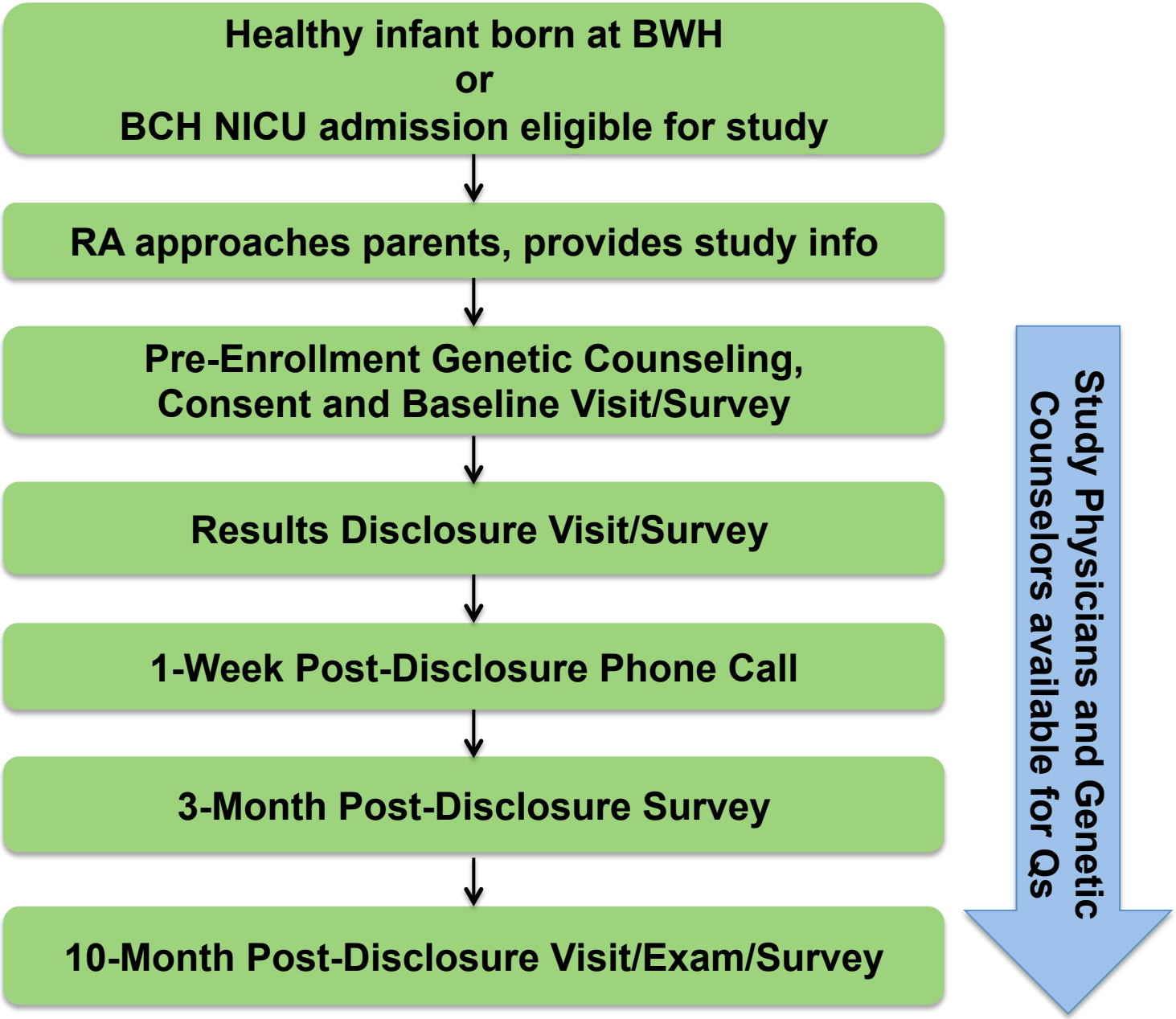
The BabySeq Project
(U19 HD077671)

- 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

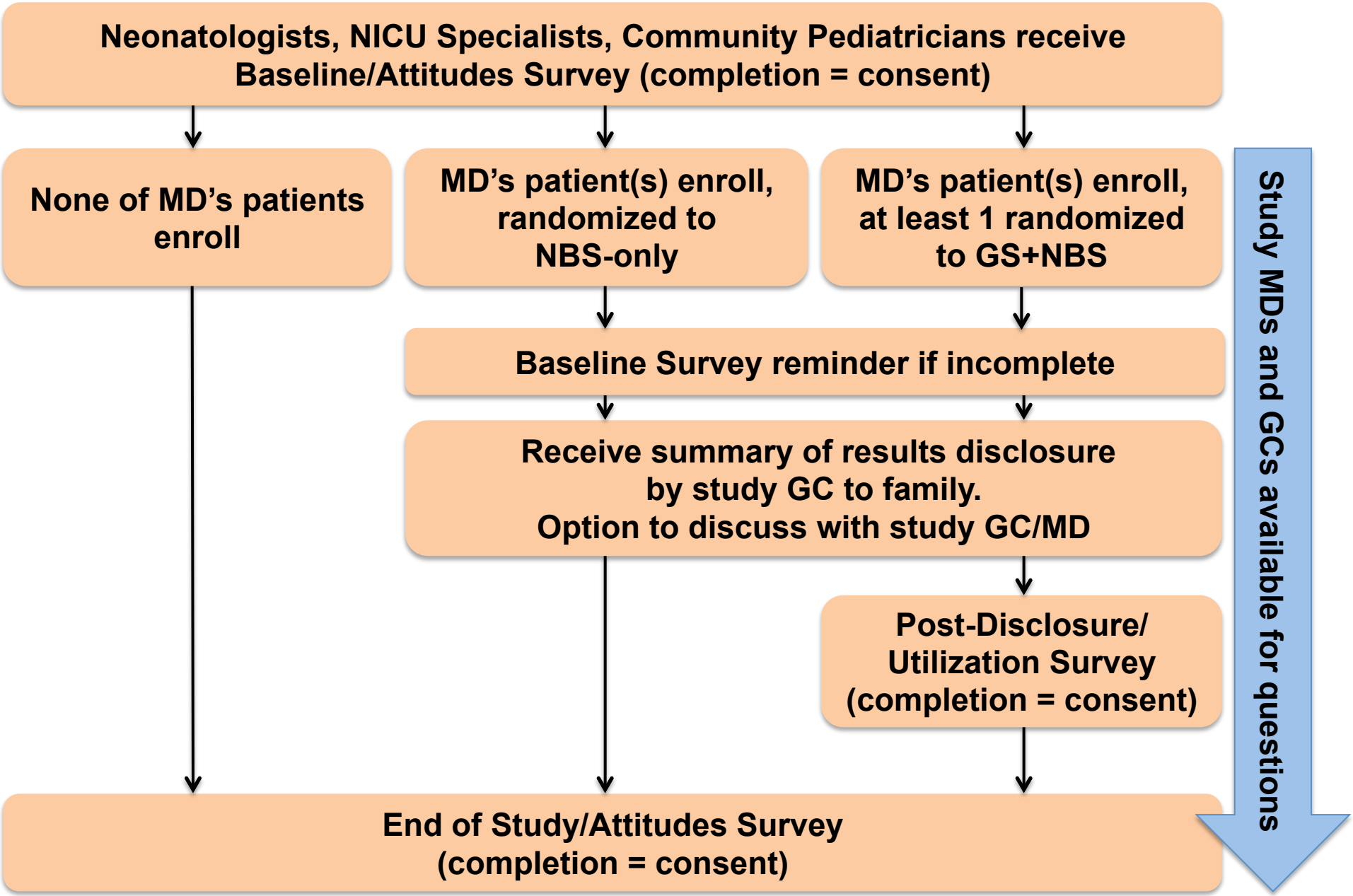
BabySeq Project Overview



Workflow for Infant and Parents



Workflow for Physicians



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

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Robert C. Green, MD, MPH (Joint PI)
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Tim W. Yu, MD, PhD
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Thank You

Questions?

Comments?

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