



BABYSEQ™

The BabySeq Project
NSIGHT Meeting
November 18, 2015



BRIGHAM AND
WOMEN'S HOSPITAL



HARVARD
MEDICAL SCHOOL

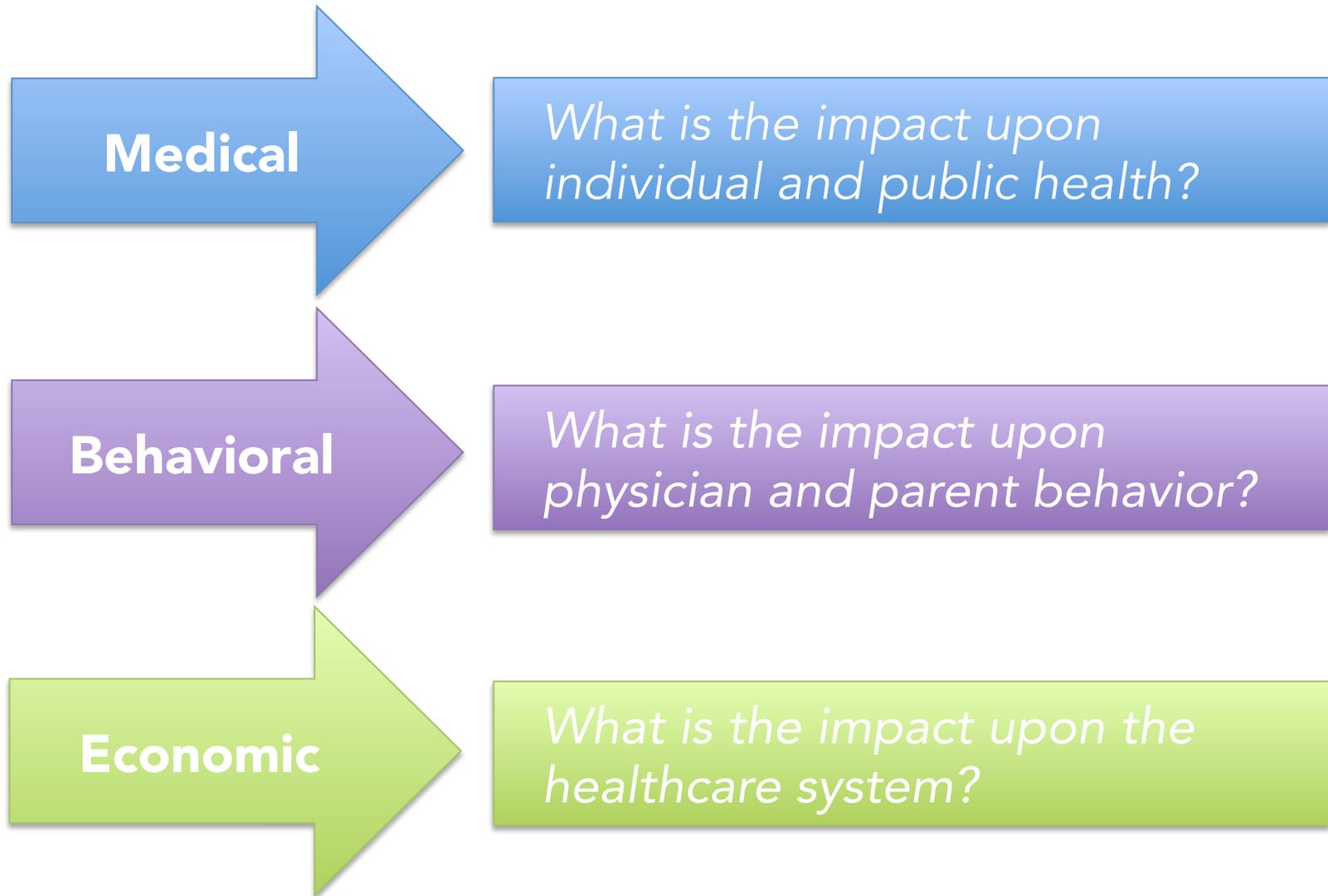
Baylor
College of
Medicine

What is the BabySeq Project?

- Research study asking “should genomic sequencing be performed on newborns?”
- Half the families enrolled in the study receive genomic sequencing
- Families and doctors are surveyed on their experience and the infants are followed over time
 - Compare the infants who received sequencing to those who did not



BabySeq is exploring the impact of genomic sequencing of newborns on families & providers.



What types of results are provided to families?

Control Group

- Family history assessment
 - Family tree interpreted by a genetic counselor
- Standard newborn screening



Sequencing Group

- Family history assessment
- Standard newborn screening
- Genomic Report
 - Disease-causing variants associated with childhood-onset disorders
 - “Carrier” status variants that should not cause disease in the infant, but may have implications for the infant and other family members
 - Pharmacogenomic variants: two specific genes which affect the way certain medications are metabolized

Family Enrollment



Who are we enrolling?



Healthy newborns from the Well Newborn Nursery at Brigham and Women's Hospital

Sick newborns from the Neonatal Intensive Care Units (NICUs) at Brigham and Women's Hospital & Boston Children's Hospital

Multi-Step Consent Process

- Initial approach by Research Assistant
- Pre-consent enrollment session with Genetic Counselor
 - Explore motivations for participation
 - Discuss types of results which may be returned
 - Average time of consent: **60 minutes** [37-130 minutes]

- Developed teaching aids

Educational Module

Developed vignettes outlining possible results for parents:

- A disease that can be treated but not prevented or cured
- A disease that can be treated and cured
- A disease that cannot be prevented or cured
- A description of carrier status

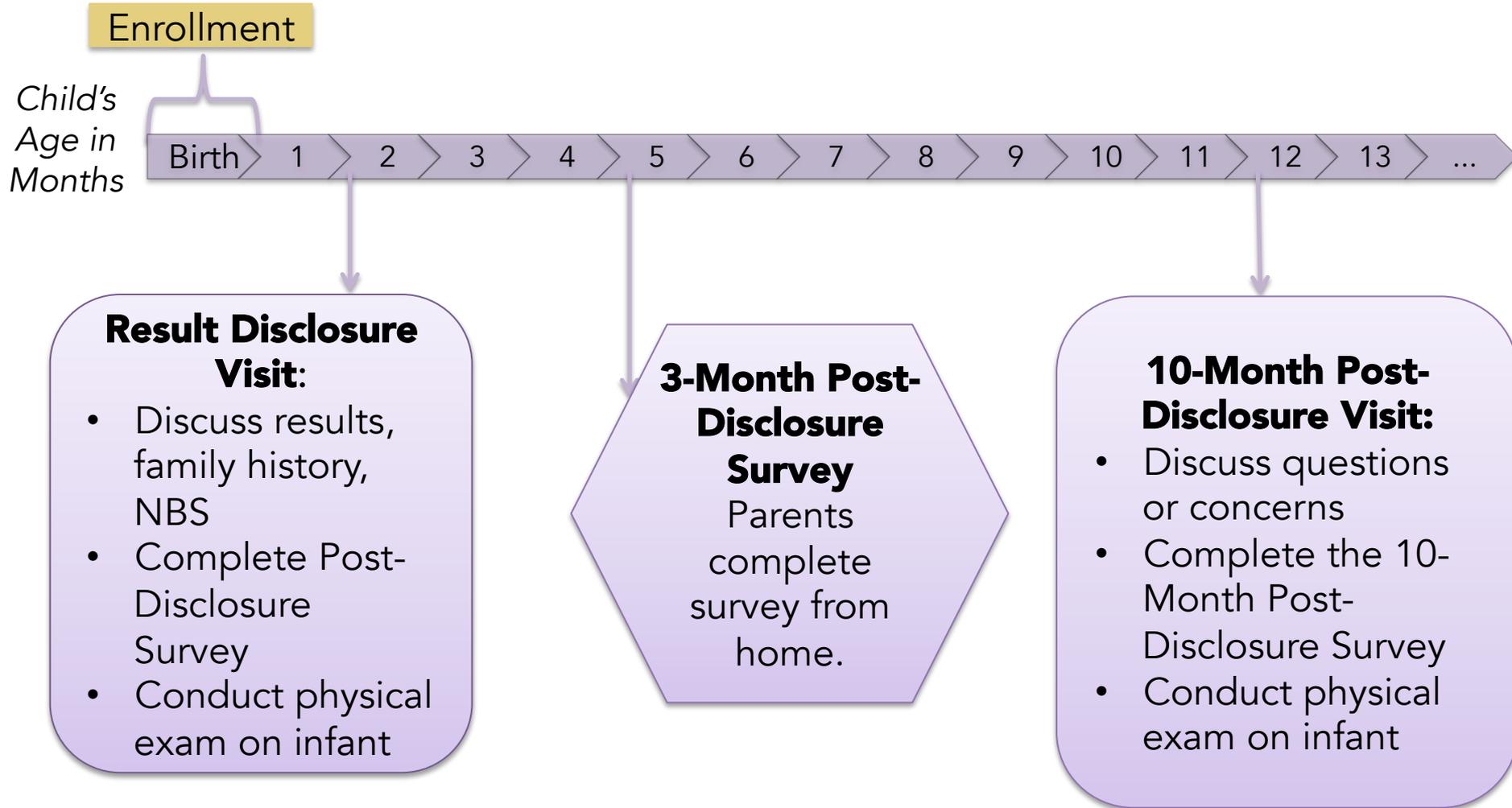
Consent Understanding Questions

18 questions that outline major components of study participation

- Any wrong answers are discussed with parents during the consent session
- Average score: **17.6/18.0** (no family has scored below 17)

- 14 day timeline after consent to complete baseline survey & finalize enrollment

Participation



Family Experiences



Family #1: “Baby Maya”

- 10 day-old infant in NICU
- Tetralogy of Fallot (TOF), a rare and complex heart condition was detected on prenatal ultrasound
- The family participated in a research study during pregnancy to study how babies with TOF develop
- As part of that study, a closure in the small intestine called duodenal atresia was detected

“Baby Maya”

- Even though “Baby Maya” needed to have her heart repaired, the intestinal problem needed to be corrected first
- She had surgery in her first week of life



“Baby Maya” Motivation to enroll

- Family was grateful to have learned about the duodenal atresia from their previous research experience
 - Allowed them to learn more about the treatment
 - Scheduled surgery, with less “surprises”
- Felt that participation in research and the opportunity to learn more could only be helpful

“Baby Maya” Study Results

- Received genomic sequencing through BabySeq study
- No cause for her birth defects were identified
- Found to be a carrier for two rare genetic changes, which will not affect her health

“Baby Maya” Impact of Results

- Day after disclosure, family saw clinical geneticist
- Genomic sequencing report was sent to clinician
- Discussion between study genetic counselor/ study lab scientist and the clinician
 - Reviewed limitations of the
 - Informed the testing protocol for the

“Excellent timing... This is very helpful”

- Clinician





Family #2: “Baby Emma”

- 18 day-old female and the first baby for the family
- Admitted to the NICU
- Birthing complications thought to cause oxygen deprivation
 - Also had a 6th finger on one hand, which was removed
 - Failed newborn hearing screen and was diagnosed with bilateral moderate to severe hearing loss
- Motivations to enroll
 - Both parents have a science background
 - Said they were “interested in research”

“Baby Emma” Study Results

- Control arm & did not receive genomic sequencing
- On the day of disclosure, “Baby Emma’s” otolaryngologist contacted a study doctor about getting results related to hearing loss genes
 - Family was in control arm → no sequencing was performed
 - However, “Baby Emma’s” DNA stored for the study could be used for the clinical testing, saving her another blood draw
 - Planned to be ordered at her next clinical appointment



Other Families' Motivations to Enroll

- Parent is adopted and not aware of health of his/her biological family
- Anonymous egg or sperm donor was used
- History of specific genetic disorder in the family
- Parent works in genetics and is interested



Conclusions

- Genomic sequencing reports from the BabySeq project can help clinicians think about a patient's diagnosis
- Genetic results have been requested for babies in the control arm
- Genomic sequencing can identify carrier status in families
- Families have many different motivations for enrolling



The BabySeq Project Team

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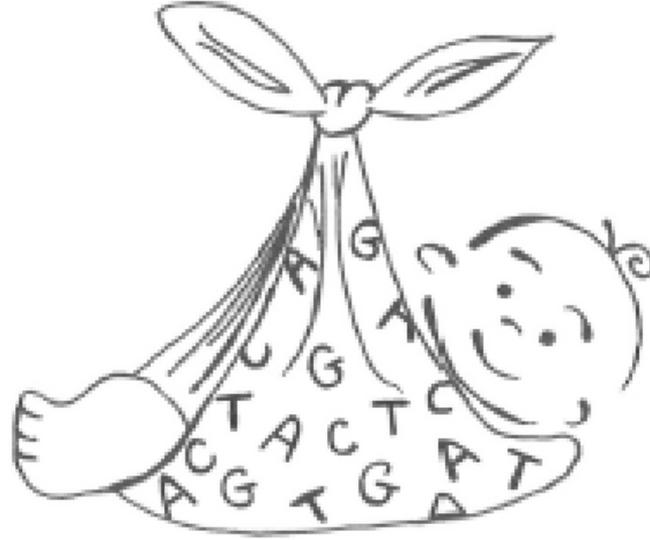
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Thank You



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