

BabySeq



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The idea of genomic sequencing for every newborn has many in the scientific community buzzing with excitement, while leaving others wary of the ethical and social implications. But what do the parents think? [The BabySeq Project](#) has been exploring parental motivations and concerns while assessing their willingness to participate in a pilot newborn sequencing study.

Led by [Alan Beggs, PhD](#), at Boston Children's Hospital and [Robert C. Green, MD, MPH](#), of Brigham and Women's Hospital, [the BabySeq Project](#) is one of four components of the NIH's [Newborn Sequencing In Genomic medicine and public Health \(NSIGHT\)](#) program. We are investigating the ethics, medical utility and economic impact of analyzing an infant's genomic information at this early stage of life – as well as parental attitudes.

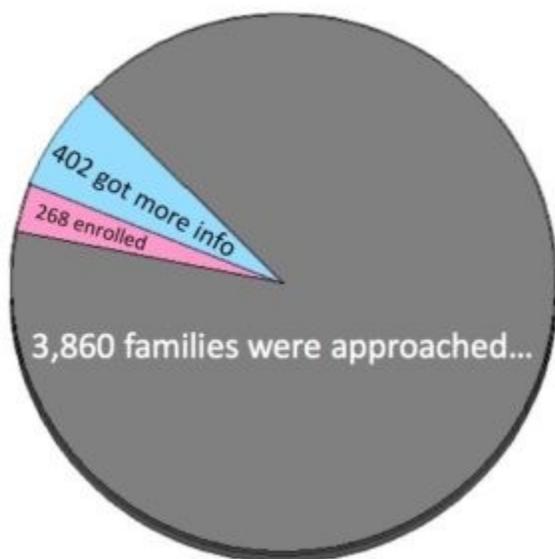
Genetic counselors like myself have had a first-hand view of the parental decision-making process. We handle much of the consenting process and talk to families in depth about what sequencing their newborn might mean.

“You want to do *what?*”

Imagine that you are a new mother, still in the hospital a mere 24 hours after giving birth. You are approached about participating in a research study that involves searching your infant’s DNA for changes in thousands of genes.

Though your baby is currently healthy, you might learn about genetic changes that indicate a likely genetic disorder in the future. Do you jump at the opportunity to learn more? Or does the thought of adding one more thing to your plate at this vulnerable moment trigger you to politely ask the visitor to leave?

If your infant is sick and hospitalized in the neonatal intensive care unit (NICU), instead of the general well baby nursery, does your instinct change?



Over the course of the study, the final enrollment rate was just over 7 percent of the 3,860 families approached. The rate was only slightly higher (about 10 percent) among parents whose babies were in the NICU. The staggering number of families who declined quickly revealed one of our most interesting pieces of data from this study: the reasons behind their refusal.

Logistical hurdles to newborn sequencing

[As we report today in *Genetics and Medicine*](#), nearly 60 percent of the “decliners” were simply not interested in research and chose not to hear any details about this study or any other. The first 48 hours of their baby’s life are filled with constant visits from providers and family. We found that our timing was a major first barrier to newborn genomic research. Who can blame parents for saying “no” to something optional in those physically and emotionally draining first days? These experiences have prompted us to considering alternative recruitment strategies in the future, such as a prenatal approach.

Even when parents were willing to consider research, we lost many because of logistics such as an extra blood draw and the need to travel back to the hospital and commit time for follow-up visits.

Initial motivations and concerns

Parents who could get past these hurdles, and whose interest was piqued at first approach, met with a genetic counselor before discharge for an education and consent session lasting nearly an hour. Our in-depth conversations were very revealing.

Many parents were drawn to the promise of personalized and preventative medicine for their child based on genetic findings, and embraced the availability of the new technology.

Yet, despite the strong interest, 1 in 3 families still opted to forgo participation. Reasons for decline at this stage were heavily focused on risks specific to genomic sequencing.

“I like more information. Knowledge is a good thing, especially if it’s actionable. Even if it’s not actionable, it’d be beneficial to know.”

“A lot of the stuff we could learn would make me really anxious, and I’m not sure there is much we can do to prevent it from happening.”

The risks (and benefits) of knowledge

Privacy and the risk of discrimination by insurance companies were strong themes. Despite federal protections provided by HIPAA and the [Genetic Information Nondiscrimination Act \(GINA\)](#), the possibility that genomic information in their child's health record might conceivably be used against them was enough to dissuade some parents from enrolling.

Other parents were less concerned about privacy, but more troubled by the potential psychological impact of learning that their brand-new, seemingly healthy baby might go on to develop a serious condition, perhaps one with no available treatment.

“[This study] gives us the ability to adapt, and I think that's more valuable than the comfort of not knowing.”

“We don't want to involve him in a medical study, without his consent, that could affect him for a lifetime. Although there are protective laws now, we don't know how this information will be used in the future.”

Others found the uncertainty of predictive genetic testing to be too extreme. The identification of a genetic variant in a newborn does not mean that they have a 100 percent chance of developing that condition. That potential for a “false positive” affected some families' willingness to be early adopters of genomic screening.

The resounding opinion of parents who enrolled was that early detection of a potential illness, while distressing, would ultimately allow their child to have the best health outcome. Others – in both the NICU and well-baby groups – felt strongly about participating in research for the benefit of the larger patient and medical community.

Not surprisingly, parents with infants in the NICU were often motivated to enroll for the chance to learn about the cause of their baby's health problem. Conversely, and perhaps predictably, NICU parents who chose *not* to enroll were more likely to cite feeling too overwhelmed with their child's health problems to commit to participation.

An ongoing dialog

Despite the hurdles and possible harms, the potential power of genomic sequencing to improve future healthcare is not lost on new parents. Several hundred couples have taken the plunge and are learning about their newborn infants' genetic findings. A small handful have learned their child carries a genetic risk for actionable health concerns [such as cardiomyopathy or cancer](#). Many parents learned that they are recessive carriers of a disease-causing variant that will not impact their infant's health, but may impact their future family planning decisions.

We are now analyzing the medical impact and parental response to genomic findings for the infants enrolled in the study and hope to report them out in the coming year. There's still much to learn about parents' attitudes toward the new technology, but this study has fueled the conversation and given us insight that will inform the growing application of genomics to the care of children.

Although this conversation is just beginning, I think we and the community at large are eager to see how this continually advancing technology will be integrated into common medical practice and its impact on patients and families.

[Read earlier coverage of the BabySeq project.](#)