Why Scientists Are Sequencing Newborns' Genomes

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(Photographer: Sandy Huffaker/Bloomberg)

Twenty years after Dolly the Sheep, the first cloned animal, was born in Edinburgh, United Kingdom, and 15 years after the first draft of the human genome was released, the dream of using DNA sequencing to prevent illnesses is

coming to fruition. Robert Green, director of the Genomes2People Research Program, explained the work of a baby genome-sequencing project at the 2016 Forbes Healthcare Summit.

Green and his team are conducting the first randomized clinical trial that sequences the DNA of newborns, dubbed the BabySeq Project. While roughly three-quarters of parents have expressed interest in knowing the genomes of their babies, few were willing to go through with the process. Of the 2,113 hospital rooms his team visited, only 7.5% of the families were willing to participate in the study.

“When you sit down with the parents, there’s a remarkable fear around privacy, around insurance discrimination,” Green explains. Lauren Stetson, who enrolled her baby daughter in the study, agrees that the process is overwhelming for new parents, and it was her husband who pushed for their participation, believing that more knowledge is always better.

By sequencing their daughter’s genes, the Stetsons found out that their baby had a biotinidase deficiency, where the body is unable to recycle the vitamin biotin. The disorder can be deadly–causing vision and hearing loss, seizures and problems with movement–but the form that Baby Stetson had was mild. As a result, a standard heel stick screening did not pick up the condition.

The Stetsons now give their daughter a dietary supplement to combat the biotin deficiency, and the baby is doing well. Another newborn in the program was found to have a BRCA2 gene mutation–a mutation that increases the risk of ovarian and breast cancer. After DNA testing, the mother found out that she also carried the same gene–a fact that she was unaware of.

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The BabySeq Project is still in its infancy, but Green thinks sequencing the genes of newborns can lead to much better preventative care and personalized medicine down the line. “We want to move from a reactive healthcare system to a proactive healthcare system,” says Green.