Would You Want to Know If Your Baby Will Get a Deadly Disease? New Study Tests Newborns' DNA for More Than 1,600 Hereditary Conditions

Courtesy Santos Family

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When Emily Santos was born last July, her parents marveled over her nearly full head of black hair and hazel eyes.

She was a healthy little girl so they didn't think too much about what may be going on inside of her – until some researchers stopped by to see if they'd be interested in getting her DNA analyzed as part of a first-of-its kind study at their hospital.

"Emily was just a few hours old and they were talking to us about the purpose of this study and how it was to help raise awareness to parents about the types of diseases or genetic disorders that a child might have," says Jaime Santos, 34, a Boston attorney who lives in Brookline, Massachusetts with her husband, Glenn, and three children.

They quickly decided to sign up.

"We were really interested in it because we're really big fans of science and medicine and trying to advance medicine," she says.

She had another reason as well.

"I actually have a close friend whose daughter has a very rare but serious genetic disorder and I remembered hearing from her about all of the challenges she had in the first year of her daughter's life trying to get it diagnosed," she says.

Emily Santos became one of the first babies to have their genomes sequenced for the BabySeqProject, a new study

assessing the risks and benefits of genomic sequencing at birth. Her parents are the first to speak publicly about their results.

The goal is to enroll about 240 healthy newborns at Brigham and Women's Hospital in Boston and another 240 newborns from the neonatal intensive care unit and Boston Children's Hospital. So far they've enrolled 100, says Robert Green, a geneticist at Brigham who is heading the study with Alan Beggs at Boston Children's Hospital.

"We hope to help the field develop appropriate, responsible ways to sequence both sick and well infants," Green tells PEOPLE. "And we hope to better understand the benefits and harms of using this technology and the cost-effectiveness in both sick infants and healthy newborns."

Half of those enrolled will get the testing and half will not. They are checking for more than 1,600 hereditary conditions that could affect a child – everything from eye cancer to rare brain diseases, Green says. They are not giving the results for any adult-onset diseases they may find, he says.

Ethical Concerns

The new study has stirred up some controversy.

"I think doing research of a genetic basis of childhood diseases makes sense but there are some very big risks involved," University of New York biomedical ethicist Arthur Caplan tells

PEOPLE.

“For one, you’re finding out things about a child,” he says. “You’re also finding out things about their biological sibling or future children that might come from the same parents. It may be more than you want to know. Or maybe more than your sister wanted to know but now they know there’s something in the family and wondering, ‘Do I have to get a genetic test to see if my kids are ruled out because my sister’s kids are ruled in?’ ”

Plus, he says, “a lot of things they find there’s nothing you can do about them. There’s no treatment. There’s no nothing. If you find out you’re at a higher risk of x then the parents just worry about it but there’s nothing they can do about it.”

Green acknowledges the controversial aspect of what he’s doing.

“This is really pushing the envelope here,” he says. “There are other groups that are doing some small-scale sequencing of newborns, particularly if those newborns have illnesses, but a large-scale sequencing of newborns is really controversial because people are concerned one may communicate to families information that is distressing, that may not have a very clear interpretation.

“So for example suppose you communicate a risk for a childhood cancer but you really don’t know how likely it is and you really don’t know what to tell the parents to do about it,” he says. “There’s no precedent for how to deal with risk information that

comes from an unanticipated screening with a genome.

The debate over sequencing is "particularly acute in infants because they can't choose for themselves," he says. "There are some ethicists who feel we should be very, very careful about assuming the baby wants that knowledge or just because the parents want it that we should give it to he family if the baby hasn't had a right to grow up and choose it.

"So that's why we're being particularly cautious about communicating adult-onset information because the baby doesn t have the right to choose or decline," he says.

Getting the Results

Jaime and Glenn discussed the pros and cons of the testing with each other then met with a genetic counselor, which is part of the study as well.

They say they were prepared for whatever the results would be – even something devastating like an incurable disease.

"If I knew there was no cure and no treatment and say it shortens the life expectancy of my child significantly I would want to know that earlier than later so I could savor even more of the time I had with her," she says.

Her husband agrees – but admits waiting for the results was a bit nerve wracking.

“I guess I was torn between thinking we’d find out everything’s great or worrying we’d find out something that would actually stress us out down the road,” says Glenn, 39, a stay-at-home dad to Emily and their nearly three-year-old twins, Sydney and Lily. “So it was pretty much in the middle.”

Emily Santos ended up in the group of infants that did get her genomes sequenced. Her parents got the report last November, about four months after the initial tests.

There were a couple of interesting things in it – like that Emily is a carrier for a disease that affects how you process cholesterol – but nothing alarming, they say.

The couple knows little about the history of their extended family so it was a relief on many levels, they say.

“When you hear things about disease or genetic defects it gets you a little nervous,” says Glenn.

They are grateful they did the testing for another reason as well. When Emily was a few weeks old, a giant birthmark appeared on the back of her left leg. Their pediatrician was concerned it could be the beginning of a genetic disease called neurofibromatosis so, after she got the report, Jaime called the BabySeq project.

“They said that they did check for that particular gene mutation and they didn’t find anything,” she says. “Now they’re going back to look at the genetic sample they took to see if they can look even more closely at that particular gene to make sure they didn’t miss anything.”

Today, Emily is a happy and healthy nine-month old and, six months after they got their results, Jaime and Glenn are still glad they did it.

“I was happy to find out there was nothing major,” she says, “and also I was able to provide that information to my brother who has children and Glenn could provide that to his siblings as well.”