MedicalXpress

Two-day test can spot gene diseases in newborns (Update)

October 3 2012, by Lauran Neergaard

Too often, newborns die of genetic diseases before doctors even know what is to blame. Now scientists have found a way to decode those babies' DNA in just days instead of weeks, moving gene-mapping closer to routine medical care.

The idea: Combine faster gene-analyzing machinery with new computer software that, at the push of a few buttons, uses a baby's symptoms to zero in on the most suspicious mutations. The hope would be to start treatment earlier, or avoid futile care for lethal illnesses.

Wednesday's study is a tentative first step: Researchers at Children's Mercy Hospital in Kansas City, Missouri, mapped the DNA of just five children, and the study wasn't done in time to help most of them.

But the hospital finds the results promising enough that by year's end, it plans to begin routine gene-mapping in its neonatal intensive care unit—and may offer testing for babies elsewhere, too—while further studies continue, said Dr. Stephen Kingsmore, director of the pediatric genome center at Children's Mercy.

"For the first time, we can actually deliver genome information in time to make a difference," predicted Kingsmore, whose team reported the method in the journal *Science Translational Medicine*.

Even if the diagnosis is a lethal disease, "the family will at least have an answer. They won't have false hope," he added.

More than 20 percent of infant deaths are due to a birth defect or genetic diseases, the kind caused by a problem with a single gene. While there are thousands of such diseases—from Tay-Sachs to the lesser known Pompe disease, standard newborn screening tests detect only a few of them. And once a baby shows symptoms, fast diagnosis becomes crucial.

Sequencing whole genomes—all of a person's DNA—can help when it is not clear what gene to suspect. But so far it has been used mainly for research, in part because it takes four to six weeks to complete and is very expensive.

Wednesday, researchers reported that the new process for whole-genome sequencing can take just 50 hours—half that time to perform the decoding from a drop of the baby's blood, and the rest to analyze which of the DNA variations uncovered can explain the child's condition.

That's an estimate: The study counted only the time the blood was being decoded or analyzed, not the days needed to ship the blood to Essex, England, home of a speedy new DNA decoding machine made by Illumina, Inc.—or to ship back the results for Children's Mercy's computer program to analyze. Kingsmore said the hospital is awaiting arrival of its own decoder, when 50 hours should become the true start-to-finish time.

Specialists not involved with the study said it signals the long-promised usefulness of gene-mapping to real-world medicine finally is close.

"Genomic sequencing like this is very practical and very real now," said Dr. Arthur Beaudet of the Baylor College of Medicine, which also is working to expand genomic testing in children. "Fast forward a year, and I think this kind of thing will probably be pretty routine."

Kingsmore said the speedy test should cost $13,500, and more study is

needed for insurers to cover it. But keeping a newborn in ICU costs $8,000 a day, and one question is if the rapid gene-mapping could shorten those stays or avoid futile care, he said.

Among the babies tested was one born with his organs on the wrong side of his body and needing emergency heart surgery. His parents had been told that it was a fluke that his older brother was born the same way, but the new test found an inherited genetic culprit that Kingsmore said will help doctors predict both boys' future treatment needs.

Three other newborns in the study died and the new test uncovered the cause of death for two of them. Kingsmore said that allowed researchers to tell parents that nothing they did during pregnancy was to blame, and to counsel them about the risks of future pregnancies.

After the study concluded, the team has performed rapid gene-mapping with additional families. It uncovered the cause of a mother's two stillbirths, allowing for assisted reproduction to help her next pregnancy be healthy, said Children's Mercy laboratory director Dr. Carol Saunders.

Copyright 2012 The Associated Press. All rights reserved. This material may not be published, broadcast, rewritten or redistributed.

Citation: Two-day test can spot gene diseases in newborns (Update) (2012, October 3) retrieved 24 April 2024 from
https://medicalxpress.com/news/2012-10-two-day-gene-diseases-newborns.html

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.