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Genetic testing for newborns

By ACSH Staff — October 4, 2012

Just last month, a series of new articles reported on the exciting findings from the ENCODE project, which is still contributing to our understanding of the human genome 10 years after it was sequenced. Now a new article published [1] in *Science Translational Medicine* is demonstrating how the genome sequencing technology can be effectively applied to infants as well as adults.

This new genetic test [2] can be used to identify diseases in newborns by scanning their DNA to look for genetic aberrations. The whole analysis takes just over two days and can provide doctors with key insight into disease diagnosis and treatment.

Approximately one in 20 babies in newborn intensive care units suffer from a genetic disease, so this new method is a good step in the right direction, according to Dr. Joe Grey, an expert in genome analysis at Oregon Health and Science University who was not involved in the study.

Although the genetic test must undergo more research before it can be used widely, the test has the potential to save many lives. All too often doctors cannot accurately diagnose genetic diseases in infants due to the complexity and sheer size of the human genome.

To determine the test s accuracy, investigators first used it to confirm a diagnosis already determined through autopsy on two babies. Next, they tested the method on four babies who were seriously ill due to a suspected genetic disease. The test quickly found the mutated genes in three of the four newborns.

This new method is quite technologically sophisticated since in addition to scanning the baby s genome, it is also equipped with a computer program that looks at specific genes based on the baby s symptoms. The test, however, solely focuses on investigating the genes that can cause diseases in newborns. For example, when sequencing and analyzing the entire genome, researchers may discover other aberrations that lead to conditions only occurring in adults, such as breast cancer. This test deliberately avoids such incidental findings.

While at this time this test seems like science fiction, given the progress in genetic fingerprinting it is to be hoped that it will become more widely available quickly, observed Dr. Ross.
