Rapid whole-genome sequencing of neonatal ICU patients is useful and cost-effective

October 19 2017

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Rapid whole-genome sequencing (WGS) of acutely ill neonatal intensive care unit (NICU) patients in the first few days of life yields clinically useful diagnoses in many cases, and results in lower aggregate costs than the current standard of care, according to findings presented at the

American Society of Human Genetics (ASHG) 2017 Annual Meeting in Orlando, Fla.

Shimul Chowdhury, PhD, FACMG, Clinical Laboratory Director at the Rady Children's Institute for Genomic Medicine, and his colleagues focused their analysis on a broad swath of NICU patients for whom a genetic diagnosis might help inform treatment decisions and disease management. They studied the clinical utility and cost-effectiveness of sequencing infants and their parents.

"Newborns often don't fit traditional methods of diagnosis, as they may present with non-specific symptoms or display different signs from older children," said Dr. Chowdhury. In many such cases, he explained, sequencing can pinpoint the cause of illness, yielding a diagnosis that allows doctors to modify inpatient treatment and resulting in dramatically improved medical outcomes in both the short and long term.

Because of the potential for early intervention and immediate adjustment in care, the researchers used a rapid WGS procedure that took three to seven days from sample collection to delivering results to patients' families. The process can be further accelerated if medically necessary. In contrast, most clinical diagnostic tests take four to six weeks.

In 34 (35%) of the 98 patients enrolled in the study, WGS yielded a genetic diagnosis, and in 28 (80%) of those patients, that diagnosis led to changes in medical management, such as the use of medications targeted to the underlying disease, avoidance of unnecessary surgery, and guidance about palliative care. Cost-effectiveness analyses are ongoing, but among the first 42 infants sequenced, the researchers calculated a $1.3 million net cost savings for that hospitalization versus the current standard of care.

"The cost savings were especially striking, given that sequencing costs are still high - even with those costs, we found that rapid WGS was not just clinically useful but economically prudent," Dr. Chowdhury said. "Given these benefits, we'd eventually like to see rapid WGS as a reimbursable first-tier test for a proportion of infants in the NICU."

Currently, the researchers are looking to expand their study and assess the effectiveness of their approach across health systems and populations. This summer, they launched partnerships with children's hospitals in California and Minnesota, an effort that will involve scaling up the rapid WGS process to meet demand and yield new insights about its clinical utility, cost-effectiveness, and ease of implementation in different environments.

Dr. Chowdhury noted the important contribution of genetics research to their progress so far. "Translational research leading to improvements in the speed and accuracy of sequencing tests is so important to our work, and has a real impact on patients and their families," he said.

More information: Chowdhury S et al. (2017 Oct 19). Abstract: Clinical utility and cost effectiveness of rapid whole genome sequencing in the neonatal and pediatric intensive care unit. Presented at the American Society of Human Genetics 2017 Annual Meeting. Orlando, Florida. ep70.eventpilotadmin.com/web/p … =ASHG17&id=170122875

Provided by American Society of Human Genetics