Clinical exome sequencing useful for critically ill infants

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(HealthDay)—Clinical exome sequencing is an effective diagnostic tool for infants suspected of having monogenic disorders, according to a study published online Oct. 2 in *JAMA Pediatrics*.

Linyan Meng, Ph.D., from the Baylor College of Medicine in Houston, and colleagues performed clinical exome sequencing for 278 unrelated critically ill infants within the first 100 days of life during a five-year period. Types of exome sequencing included proband exome, trio exome, and critical trio exome, which is a rapid genomic assay for seriously ill infants.

The researchers found that a range of medical concerns were included as clinical indications for exome sequencing. Clinical exome sequencing achieved a molecular diagnosis in 36.7 percent of infants, with the yield for cardiovascular abnormalities relatively low. For 52 percent of infants, the diagnosis affected medical management, with a considerable impact on informed redirection of care, initiation of new subspecialist care, modifications to medication/diet, and furthering lifesaving procedures. A molecular diagnosis was revealed in 50.8 percent of infants undergoing critical trio exome sequencing; patients who underwent critical trio exome sequencing had significantly different diagnostic yield, patient age at diagnosis, and medical effect compared with those who underwent regular exome testing.

"Exome sequencing is a powerful tool for the diagnostic evaluation of critically ill infants with suspected monogenic disorders in the neonatal and pediatric intensive care units and its use has a notable effect on clinical decision making," the authors write.

The Department of Molecular and Human Genetics at the Baylor College of Medicine derives revenue from the clinical exome sequencing offered at Baylor Genetics. One author disclosed ties to Veritas Genetics China.

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