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Genetic Testing of Critically Ill Children Can Identify Disease, Guide Treatment

Nearly two-thirds tested in pediatric intensive care unit found to have link to genetic condition, study shows

- Rapid genetic testing of critically ill children can help clinicians identify the problem and change the course of care: study presented at SCCM's 2023 Critical Care Congress.
- Nearly two-thirds of children tested in the pediatric intensive care unit (PICU) had a genetic variant.
- Among the children who had a positive genetic test, more than a third underwent a change in their care, such as a medication or procedure.

SAN FRANCISCO – Genetic testing of critically ill children can help healthcare professionals diagnose their condition and improve outcomes, suggests a study presented at the Society of Critical Care Medicine's (SCCM) 2023 Critical Care Congress.

In this first multicenter study to investigate the potential benefits of rapid whole genome sequencing (rWGS) in the PICU, a genetic variant tied to a medical condition was identified in nearly two-thirds of children tested. More than one-third of the children were found to have a genetic cause of illness that led to a change in management. The most common genetic causes were linked to heart and neurologic diseases.

"These findings suggest that rWGS is a powerful addition to care in critically ill children in whom a genetic cause is suspected or the cause of their illness is unclear," said Katherine Rodriguez, MD, lead author of the study and a pediatric critical care fellow at Rady Children's Hospital in San Diego. "While rWGS is becoming standard in the NICU, it is still rare in the PICU; these findings suggest it can be beneficial and help alter the course of treatment for some children." Infants who are severely ill when they are born are placed in the neonatal ICU (NICU). The PICU is designed for critically ill children up to age 21.

The study was conducted at three children's hospitals from March 2019 to July 2022. A total of 80 children whose cause of illness was unclear underwent rWGS testing, with 65% testing positive for a genetic variant. Among children with congenital heart disease or sudden cardiac arrest, 72% had a genetic diagnosis and 50% of those with a suspected neurologic disease had a genetic diagnosis.

Among the children with a genetic diagnosis, 38% underwent a change in management: 21% while they were in the PICU and 34% during their hospital stay after leaving the PICU. Changes included medication (81%) or procedure (56%), such as avoiding a biopsy or having surgery.

For example, a physician was unsure whether a child's heart condition was caused by an infection or a genetic cause and performed rWGS, determining within 48 hours that it was the latter. The child was quickly listed for—and soon received—a heart transplant without needing to wait for the infectious disease test results. Another child with severe respiratory failure and fluid in the lungs tested positive for a rare immune disorder and received additional immunizations to avoid another infection.

"Often these children don't have obvious features or developmental delays associated with a syndrome," Dr. Rodriguez said. "Candidates for testing include children who are very sick or have an unusually severe course of illness and should have improved faster. PICU clinicians who are feeling like something is not right with a patient should consider genetic testing."

THE SOCIETY OF CRITICAL CARE MEDICINE

The Society of Critical Care Medicine (SCCM) is the largest nonprofit medical organization dedicated to promoting excellence and consistency in the practice of critical care. With members in more than 100 countries, SCCM is the only organization that represents all professional components of the critical care team. SCCM's Critical Care Congress brings together intensivists and critical care experts from around the world to share the latest scientific research, develop solutions to common issues, and improve the care of critically ill and injured patients. Visit sccm.org for more information. Follow @SCCM or visit SCCM on Facebook.

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