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Rapid Genomic Sequencing in NICU Patients-Past, Present and Future

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Background: The effectiveness of targeted gene panels to improve the diagnosis and care of critically ill infants remains unknown. Methods: After developing RapSeq, a panel targeting 4,503 disease-causing genes, we selected candidate patients for testing in our neonatal and pediatric/cardiac intensive care units (NICU and PICU/CICU) based on specific criteria. Fifity infants and their parents were sequenced from October 2018 to March 2021. We assessed diagnostic yield, turnaround times, and clinical consequences; and we performed cost analysis of one case. Results: a firm diagnosis was made in 25/50 neonates (50%); thirteen had de novo variants, eleven were either compound heterozygote or homozygous, and one had a maternally inherited GNAS variant. Preliminary reports were generated by 9.6 days (mean); final reports after Sanger sequencing at 16.3 days (mean), with no discrepancies. In all positive infants, the diagnosis changed management. In two cases with congenital myasthenia, diagnosis and treatment occurred at 17 days with RapSeq vs. 7 months, and hospitalization costs were at least $71,393 higher with conventional testing. Conclusion: This study shows that a gene panel that includes the majority of known disease-causing genes can rapidly identify a diagnosis in a large number of tested infants. Due to simpler deployment, easier variant interpretation, and lower costs, this approach might represent a valid alternative to exome and genome sequencing in the care of critically ill newborns.

Biography

Dr. Mao is the medical director of Molecular Genetics and Genomics in ARUP Laboratories and a professor of pathology at the University of Utah School of Medicine. She received her MD from Capital University of Medicine in Beijing and her MS in molecular pathology from Beijing Union Medical College. She is board certified by the American Board of Medical Genetics, with a subspecialty in clinical molecular genetics, and certified with the New York State Department of Health, with a subspecialty in genetic testing. Her research interests include the genotype-phenotype correlations in inborn errors of metabolism and genetic diseases in the RAS/MAPK pathway; she is also involved with implementing next-generation sequencing techniques into molecular diagnostics.