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Gene Technology

Commentary

Impact of Genetic Medicine in Providing Access to New Borns in Neonatal Intensive Care

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DESCRIPTION

Genomic Medicine (GM) has quickly advanced as exome sequencing was first utilized for patient diagnosis in 2010. The most likely population to benefit from GM is probably critically unwell infants admitted to Neonatal Intensive Care Units (NICUs), who have high rates of genetic abnormalities and associated morbidity and mortality. This is because early diagnosis is essential to maximize the advantages of early tailored management, which includes a growing number of cutting-edge personalised therapies. Many studies have shown the diagnostic, therapeutic, psychological, and educational value of Genomic Sequencing (GS) for critically ill children with underlying genetic abnormalities and their families. GS has the ability to change neonatal treatment. To promote equal access in this crucial group, solutions are urgently needed because it is arguable that the current use of GM in clinical treatment has aggravated health inequities.

Infant mortality and substantial morbidity caused by genetic abnormalities are often lifelong effects. The major causes of infant mortality in the United States are now thought to be genetic, to advancements in obstetric care and neonatology over the past several decades that have decreased morbidity and mortality from other perinatal disorders. Precision medical approaches will be needed to reduce M and M caused by genetic illnesses, and the first step in this process is to uncover the underlying genetic diagnoses. Due to limited access to genetic testing, up to 25% of severely ill infants in NICUs may have an undetected genetic disease. In general, any infant who is seriously unwell and has a condition may be suspected of having an underlying genetic abnormality and be eligible for GS. Many congenital defects, as well as neurologic, metabolic, or other serious organ systems, may be included in the list of particular phenotypic criteria in the neonatal period to prioritise for GS, according to our personal experience. Infants with one or more congenital abnormalities are advised to receive GS, according to a recent evidence-based recommendation from the American College of Medical Genetics and Genomics.

For critically unwell children with suspected genetic abnormalities and their families, rapid GS presents a variety of potential advantages. Initially, GS can result in a genetic diagnosis and put an end to the diagnostic journey. A weighted average diagnosis rate of 36% was recently published in a review of 31 trials on fast GS in new born and paediatric patients in intensive care settings. The same review found a weighted average change in management rate of 27%, suggesting that GS may result in a change in clinical management. A genetic diagnosis may affect the course of treatment and the intended outcomes. An additional benefit of a genetic diagnosis is that it may open doors to specific studies like natural history investigations and clinical trials of cutting-edge precision treatments. We further point out that nondiagnostic GS has occasionally been documented to result in management changes. Finally, prognosis and reproductive counselling may be affected by a genetic diagnosis for example, the likelihood of intellectual disability or developmental delay for the former and the risk of recurrence for the latter. Fourth, extra patient-reported advantages for the new born and family may result from a genetic diagnosis. Finally, we want to mention that recent shown how economical GS is in NICU settings. Their view is that GS now has the most potential to influence the medical care of the NICU population, even while we acknowledge that access to genomic medicine is uneven across our health care system. In this viewpoint, we describe hurdles to fair access to genetic medicine and suggestions to eliminate those barriers using critically sick infants in NICUs as a paradigm population.

Implementation of genomic medicine

There are numerous points in the GM implementation process where barriers to equitable access exist. It is crucial to understand that racism on the internalised, interpersonal, institutional, and structural levels may be a barrier at each step. Insufficient knowledge of genetics and genomics, difficulty correctly recognizing infants with suspected genetic abnormalities, and/or ambivalent attitudes about genetic testing may all be problems for healthcare professionals without formal training in the clinical

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genetics. The next step is for GS to be authorised and clinically accessible. While neonatal intensive care is provided in a variety of locations, GS is mostly offered at big academic referral facilities that have the tools and know-how to carry out this process sustainably. Where GS is not available, infants in community and/or rural NICUs may have minimal or no genetic testing, or they may be moved to referral centres for a thorough genetic analysis, which may place additional responsibilities on the families and increase expenses on the healthcare system. A provider must give the baby's family pre-test counselling and obtain agreement before ordering GS. Where accessible, clinical geneticists or GCs frequently perform this task, therefore Non-Genetics Providers (NGPs) might find it uncomfortable. Furthermore, due to institutional racism and the historical injustices, as well as additional challenges including language, literacy, and cultural differences, families of racial and ethnic minorities may be more prone to lack confidence in genetic testing and/or the healthcare system. Samples must be gathered and sent to the sequencing lab with the family's permission. The collection of the new born sample is very simple in the NICU, but the collection of parental samples, which enhance the interpretation of the data, may be more difficult for families that have access to limited means of transportation, child care, employment, or other factors. Third, families from marginalized groups may find it challenging to obtain clinical geneticists and other subspecialists for complex follow up after NICU release due to the prevalence of discovered genetic disorders.