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Gene Technology

Commentary

Genetic Disorders and its Mortality in Infants

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DESCRIPTION

In Neonatal Intensive Care Units (NICUs), infants with genetic disorders, such as chromosomal abnormalities (aneuploidy syndromes or chromosomal deletion or duplication disorders) are monogenic. Mendelian disorders (caused by variants in single genes), contribute significantly to mortality. Major congenital malformations, which impact about 2% of all births. And also the main cause of infant mortality though the underlying a etiology of these deformities may or may not be genetic [1]. The combined contributions of genetic disorders and congenital malformations to neonatal and infant mortality have previously been estimated at 20–50% depending on the population studied, though these figures were often calculated in the absence of comprehensive genetic testing and relied on clinical diagnoses.

Because of the growing use of massively parallel sequencing techniques, particularly Exome Sequencing (ES), there is still a greater awareness of monogenic conditions that present in infancy, it is likely that the known contribution of genetic disorders to infant mortality will continue to rise [2]. Adequate efforts to reduce infant mortality cannot be achieved without adequately acknowledging the significance of genetic disorders, particularly mendelian disorders. We wanted to know more about the genetic evaluation and diagnostic yield of genetic testing in situations of newborn mortality. As massively parallel sequencing technologies for diagnosis, particularly ES and GS, become more widely used in the neonatal or infant period and have demonstrated high diagnostic utility it is likely that our understanding of the phenotypic spectra for many mendelian disorders will continue to expand, and more mendelian disorders will be recognized as contributors to infant mortality [3]. As a result, the growing usage of ES and GS is anticipated to have an impact on the landscape of genetic factors to infant mortality. Furthermore, it has been suggested that increasing the use of ES or GS in the NICU will increase newborn (under 28 days old) mortality by assisting parents in making the tough decision to transition to palliative care measures that may shorten life or cause death. To discontinue life-sustaining treatments, even if total infant mortality is reduced as a result of discovery of treatable conditions [4]. Infants with genetic diseases have a greater day mortality rate than those without a diagnosis, which could be due to the high diagnostic yield for particularly severe phenotypes or an acceleration of the period to switch care goals or discontinue life support.

As the scientific ability for genetic diagnosis improves, a growing percentage of diagnoses are made in childhood or even in the newborn. Many of these diseases have been linked to developmental delays and intellectual disabilities, which are not clinically visible at the time of diagnosis. Others may be linked to cognitive impairment, although the prevalence and severity of these conditions are unknown. As a result, these genetically diagnosed neonates and babies represent a growing population of individuals at high risk for neurodevelopmental disorders [5]. After discharge from the NICU, there are well-established developmental supports for high-risk newborns, particularly preterm infants, but programs expressly for children with genetic diagnoses are rare. Medical problems that appear in the newborn period can have a significant impact on developmental outcomes. Preterm birth and low birth weight, as well as serious congenital defects, have been linked to developmental delays and a variety of hereditary disorders, including metabolic inconsistencies. Many developmental problems, such as autism spectrum disorders, and neurosensory disorders, such as hearing loss, tend to benefit from earlier detection and treatment.

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