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NEWBORN GENOME-SEQUENCING UNLOCKS THE BLUEPRINT OF HEALTH

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Representative image: A technician prepares to sequence genetic material at a research facility. | Photo Credit: nci/Unsplash

This article is part of a fortnightly column exploring contemporary concepts and issues in genetics.

Imagine a situation where a severely ill newborn is in the ICU and a fast, effective diagnosis could enable effective treatment – a scenario that plays out practically in every neonatal ICU on a regular basis. The situation is complicated when the disease affecting the baby is not common and known to many clinicians, and could be buried in medical textbooks or databases.

There are 6,000 or so genetic diseases, of which around 3,500 diseases have been documented, and a much smaller number have had their molecular and/or genetic defects mapped.

A significant number of diseases in the population are also treatable but are nevertheless prevalent.

Newborn screening programmes now in vogue in different countries, and which have been deployed in some states in India as well, are based on the fact that an early diagnosis could allow us to use effective treatments and save an infant from death or disability.

For example, in the U.S., healthcare workers screen for around 30 diseases, including treatable ones of the blood, the endocrine system, and metabolism.

Then again, in many cases, they lose the window of opportunity because standard newborn-screening programmes are limited in the menu of genetic tests they cover.

Thanks to recent advances, genomic-sequencing is now available, accessible, and in many ways more affordable. It also offers a much better coverage of genetic diseases to screen for. Importantly, this could help healthcare workers make a fast and effective diagnosis, helped by the fact that sequencing is also a ‘single’ test, versus the multitude of tests performed as part of routine newborn-screening.

The rarity of many genetic diseases, the narrow window of opportunity, the long diagnostic paths, and the unfortunate deaths of ill newborns makes it very difficult to document and understand these diseases. However, population-scale genome-sequencing efforts have provided insights into the prevalence of many of these diseases in an unbiased manner.

Discoveries in the last three decades have also allowed a small but significant number of diseases to be treated or managed effectively. This in turn opened up a newer opportunity: to diagnose and treat genetic diseases through genomic-sequencing in newborns, especially sick newborns.

Researchers at the Rady Children's Institute, led by Stephen Kingsmore, earlier showed that whole-genome sequencing could provide a much higher number of positive cases with a diagnosis, around 40% (compared to standard genetic tests at 10%), with 26% of the diagnosed children benefiting from reduced severity of illness due to the rapid diagnosis and, consequently, a significant reduction in the cost of treatments.

Another report a year later from researchers in the U.K. also reported numbers consistent with previous reports.

The benefits of sequencing may not just be limited to newborns who are unwell. The BabySeq project funded by the U.S. National Institutes of Health is one of the most comprehensive studies to evaluate sequencing of newborns for routine newborn care.

One recent study conducted by the project, and published in the *American Journal of Human Genetics*, evaluated the sequences of 127 apparently healthy and 32 sick infants. It found that just over 10% of infants had an unanticipated risk of genetic diseases. When these infants were followed up for three to five years, sequences revealed the causes of disease in three infants; in the remaining 14, a better picture of the risk made way for better medical surveillance.

The sequencing also warranted additional at-risk family members of 13 infants to have their genes sequenced. Three of them benefited from subsequent surgeries.

Another recent study, published in *JAMA Network Open*, surveyed over 200 genetic experts. Most of them firmly believed that sequencing newborns should be part of routine care.

So it isn't surprising that the U.K. National Health Services recently launched a nationwide programme to sequence 100,000 sick newborns.

The fight for who can sequence the fastest started with the first Guinness Book of records entry: in 26 hours, by Dr. Kingsmore & co., a mark his team broke in 2018 for a time of 19.5 hours. In 2021, Euan Ashley and team got there in just over 5 hours and 2 minutes

Records apart, a large study with more than 100 children with different disease complexities, and published in 2019, suggested a median time for sequencing, clinical interpretation and reporting of just over 20 hours, suggesting the approach could have far-reaching impact in clinical settings.

With technological advances, including better AI-based tools to assist clinical decisions, rapid sequencing is likely to become a diagnostic mainstay for unwell infants in clinics.

Newborn whole genome sequencing presents multiple ethical challenges. First: the issue of disclosing and managing incidental and secondary findings raises concerns about privacy and the psychological impact on families. Updated recommendations by the American College of

Medical Genetics and Genomics regarding secondary findings could help deal with incidental findings.

The equitable distribution of benefits and burdens associated with accessing and utilising this technology also invoke issues of justice and fairness .

As the vast potential of rapid newborn whole-genome sequencing unfolds, we stand at a crossroads of hope and introspection. There is no doubt that this technology will help clinicians with the means to detect rare genetic disorders, anticipate susceptibility to disease, and give them the evidence required to prescribe better treatments and shape a healthier future. Yet we must also tread carefully, considering the delicate balance between benefits and harm.

If we do, then it may not be far-fetched to imagine that rapid whole-genome sequencing will be the right of every child in the years to come.

The authors are scientists at the CSIR Institute of Genomics and Integrative Biology. All opinions expressed here are personal.

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