Genome sequencing of healthy newborns

NHS England proposal

At the Genomics England Research Conference on 4th November 2019, Matt Hancock, MP, Secretary of State for Health, announced that NHS England would offer genome sequencing to healthy newborn infants, volunteered by their parents, as part of a pilot study. The intention is that this will identify children at risk of life-threatening conditions to enable them to access early treatments. He proposed that genome sequencing would be rolled out to all healthy newborns in England within three years.

BSGM response

The British Society for Genetic Medicine (BSGM) supports the use of genome sequencing as a diagnostic tool in newborn infants and children presenting with serious problems consistent with a genetic condition. In this situation, early diagnosis can be lifesaving, for example, if the infant is found to have a treatable metabolic condition. The BSGM membership- NHS doctors, scientists and genetic counsellors- already deliver such a service.

Whereas genomics can be very good at diagnosing conditions when the clinical context is taken into account, the genetic code of a healthy newborn will only rarely predict future disease accurately. Parents might be drawn to genome sequencing of their baby in the hope that it will provide clear cut information about their future health, but factors other than the readout from the genetic code are necessary to do this well. Such a venture therefore needs to be carefully researched, and the ethical and societal aspects require careful consideration before roll-out to the general population. The pilot research study will therefore need to evaluate which results will be communicated as well as what parents understand might be communicated to them. Those results that are communicated should abide by current professional advice which recommends that children are not tested for adult onset conditions if there is no effective preventative intervention or treatment in childhood. Issues such as sample and data storage, access and retrieval also require detailed scrutiny.

Before genome sequencing is rolled out into practice, it is important that the programme meets the criteria for screening set out by the National Screening Committee. These include that the pilot study gathers information on the health economic benefit of this approach, comparing it with current methods of newborn screening. The study must also seek to explore ethical concerns and consider the long-term impact on the child and their family, and address issues such as data storage and retrieval in the presence of future health concerns.

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