
**Scientists were stunned by the number of babies with unanticipated genetic findings that could lead to disease prevention in the future**

'The science is easy to process; the emotional component is not,' Dr Rajani Aatre, a genetic counselor at the University of Michigan Frankel Cardiovascular Center, **warns**.

'Let's say you find out your kid got something because you passed it down. No matter how much you intellectualize it, you can't ever discount that the feeling of responsibility or guilt won't affect you.'

The BabySeq project was set up by researchers at Brigham and Women's Hospital in Boston and Boston Children's Hospital, offering gene sequencing to the parents of healthy and sick newborns.

Robert Green, co-director of the study and a Professor at Harvard Medical School, said: 'The BabySeq Project is the first randomized trial of sequencing in newborns and the first study to fully examine the wealth of unanticipated genetic risk information in children.

'We were stunned by the number of babies with unanticipated genetic findings that could lead to disease prevention in the future.'

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Mutations linked to several heart conditions were spotted. These included six newborns with variants associated with dilated or hypertrophic cardiomyopathy. These can lead to heart failure.

Another had a version of a gene associated with supravalvular aortic stenosis - a congenital disorder that narrows the large vessel carrying blood from the heart which is also potentially fatal.

These conditions can be monitored over time - and the families have been referred to cardiac specialists.

Another baby was found to have a variant that had caused the infant vitamin B deficiency - a condition that can cause skin rash, hair loss and seizures.

The child's diet is now being supplemented with the nutrient biotin which should prevent the condition.

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As genome sequencing becomes increasingly commonplace in the clinic its use and role among newborns is controversial because of the distress it could cause families.

The findings, published in the American Journal of Human Genetics, shed light on how common it is to find something important to a child's future health - and the benefits or consequences for families.

Experts believe it could one day be part of routine testing done on every baby - providing doctors and parents with a vast pool of data.

It's likely to reveal a wider range of potential medical risks than the traditional heel-prick test.

Used across the world - including the UK - a small sample of newborns' blood is taken to check for more than two dozen possible conditions.

The BabySeq Project enrolled healthy newborns from the Brigham's well baby nursery and ill newborns from BCH, the Brigham and Massachusetts General Hospital's neonatal and paediatric intensive care units.

Family histories were collected for all participants. Half of the families from each group were randomised to receive standard care.

This included 'heel prick' screening and genetic counselling based on family history. The other half received whole DNA sequencing in addition with results disclosed to families during the counselling session.

The team monitored health outcomes and collected medical, behavioral and economic data at the time of disclosure and three months and 10 months later.

This measured the impact of genomic sequencing on the infant's clinical care, parent and clinician behaviours and economic outcomes.

A total of 159 newborns - 128 healthy newborns and 31 ill newborns - had their DNA mapped.

Of those, 15 (9.4 percent) were found to have a mutation for which increased the risk of a disorder during childhood or for which an intervention might prevent devastating outcomes later in life.

Senior author and BabySeq co-director Dr Alan Beggs, director of The Manton Center of Orphan Disease Research at Boston Children's Hospital, said: 'Sequencing results have potential to raise questions that may be upsetting for parents but could also lead to helpful or even lifesaving interventions.

'Only time will tell how the costs - both financial and in terms of extra medical testing and family stress - balance out against the benefits. That's what we're really trying to find out.'

The team also offered to provide information regarding their child's risk for medically actionable, adult onset conditions.

Three of 85 infants of parents who agreed to receive this information carried such variants. These mutations were also identified in the mothers of the three children.

Prof Green added: 'Disclosing genetic risk for adult onset conditions in children has been discouraged in traditional genetics in order to protect the child's right "not to know," but our results demonstrate that many parents want access to this information about their child.

'Our findings suggest that sequencing newborns thoroughly reveals potentially life-saving information in both infants and their parents far more commonly than was previously thought, and should encourage our entire field to re-evaluate the value of comprehensively analyzing and disclosing genomic information at any age.'


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