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With gene screening, ATLAS shrugs off parents' worries

New mother Katerina Babajanov with three-week-old Alexandra at home in Sydney. Picture: Jane Dempster

EXCLUSIVE

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A new method of screening for newborn babies is for the first time set to allow checking for multiple genetic conditions using a single test.

The advance is the result of new DNA gene sequencing technology that allows hundreds of genes to be simultaneously analysed for variations that cause genetic diseases.

The technology, known as ATLAS, has been developed by Australian company Genepath and Queensland's public pathology arm, Pathology Queensland.

The organisations collaborated on a feasibility study conducted in Australia that has provided "proof of concept" for the potential of next-generation DNA sequencing technology to dramatically expand population newborn screening by detecting disease-causing gene variants while avoiding revealing an individual's entire genetic make-up, which can result in unintended consequences.

Unlike sequencing an individual's entire genome, the ATLAS screening platform provides targeted screening, honing in on 164 genes most commonly associated with genetic diseases for which treatments are available. The results of the feasibility study are published in the journal Clinical Chemistry.

"This is really a breakthrough technology because it has the potential to really significantly expand the numbers of conditions screened," says Glenn Bennett, Genepath's chief medical officer and an adjunct fellow at the University of New South Wales School of Population Health.

"This new technology offers the chance to test multiple conditions at the same time in a single test, and really significantly improve preventive health in Australia."

Currently, newborns are given a heel-prick test, which screens for about 30 conditions.

Next-generation DNA sequencing is a technology that can determine the sequence of DNA or RNA to study genetic variation associated with diseases, which enables the sequencing of many DNA strands at the same time and can detect many hundreds of conditions simultaneously.

Sydney mother Katerina Babajanov's daughter, Alexandra, was given the heel-prick test when she was born three weeks ago. Ms Babajanov, who plans to have another baby in future, said she would welcome an expanded newborn screening program.

"I think the fact that there is an available treatment for the particular screened genetic conditions is important so that it doesn't increase parents' anxieties."

Dr Bennett said if the ATLAS screening technology was used on a population-wide basis in Australia targeting 164 genes, about 1.3 per cent of all babies screened would be expected to test positive for a specific condition, which would increase the yield of the nation's newborn screening program ten-fold.

"Whole population newborn screening is a highly effective public health intervention as it reduces death and disability from treatable genetic diseases," Dr Bennett said. "Through this feasibility project, we've shown that this technology could be implemented into newborn screening in Australia today."

Jacobus Ungerer, director of chemical pathology at Pathology Queensland, said the study had demonstrated it was feasible and practical to carry out targeted gene sequencing for newborns but pathology labs would have to upscale to implement the technology.

“Introducing targeted gene sequencing will enable the diagnosis of serious treatable genetic conditions in numerous babies,” Dr Ungerer said. “The health and economic benefits to society will be substantial.”

Genepath will carry out an implementation pilot project with Pathology Queensland next year using the technology.

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Natasha Robinson began her career at The Australian in 2004. A Walkley awards finalist and a Kennedy Awards winner, she was appointed Health Editor in 2019, and has covered rounds including national affairs, indige... Read more