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media@unsw.edu.au

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Newborn screening for spinal muscular atrophy improves chances of walking and quality of life: study

Testing in the newborn period is more effective than conventional clinical diagnosis pathways for improving health outcomes in infants with the rare genetic disease.

Children with spinal muscular atrophy (SMA) are more likely to walk, be more functionally independent and free of respiratory and feeding support when screened, diagnosed and treated shortly after birth, according to a new study conducted at Sydney Children's Hospitals Network (SCHN) by UNSW Sydney researchers.

Significantly, the findings show newborn bloodspot screening (NBS) for SMA, coupled with potential to access disease modifying therapies, is correlated with greater motor milestone acquisition with those diagnosed before the onset of symptoms reaching regular childhood developmental milestones.

The study, published in *The Lancet Child & Adolescent Health*, is one of the first to investigate the effectiveness of NBS for SMA beyond clinical trial populations.

"The research shows the effectiveness of newborn screening for spinal muscular atrophy in the broader population," says the lead author of the study Dr Didu (Sandi) Kariyawasam in the School of Clinical Medicine, UNSW Medicine & Health, and a paediatric neurologist at SCHN.

"It's an important study for building the evidence base that newborn screening is an approach that leads to better health outcomes for children with spinal muscular atrophy and is impactful enough to justify widespread adoption," says Professor Michelle Farrar in the School of Clinical Medicine, UNSW Medicine & Health, and a paediatric neurologist at SCHN.

Early diagnosis means early treatment

SMA is a childhood-onset motor neuron disease. Left untreated, it is a potentially fatal genetic condition caused by a missing or faulty *SMN1* gene, which leads to progressive muscle weakness and wasting in babies – limiting their ability to move and sometimes feed and breathe independently.

Clinicians are usually only able to diagnose and treat SMA after symptoms have appeared. By that point, many have already and irreversibly lost up to 90 per cent of their motor nerves, which is why earlier detection is vital.

"Until recently, SMA was the leading genetic cause of infant mortality worldwide. Many babies born with the severest infantile-onset form of the disease – type 1 SMA – died before their second birthday," says Dr Kariyawasam.

NBS programs test for a range of rare, treatable conditions as early as possible after birth – even before symptoms appear. While there is currently no cure for SMA, recent advances in the effectiveness of genetic treatments have opened the possibility for a test for SMA to be included in newborn bloodspot screening.

“While the development of genetic therapies has been crucial, having a proactive model for early diagnosis is equally as important as an intervention,” says Dr Kariyawasam. “The earlier the diagnosis, the earlier treatment can begin, which is where newborn screening comes in.”

Newborn screening improves health outcomes

For the study, researchers followed 15 children diagnosed with SMA using NBS (screening group) and 18 children diagnosed by clinical referral (comparator group) for two years from diagnosis. Children within both groups had the potential to access disease-modifying therapeutics.

The two-year survival rate was 93 per cent in the screening group and 89 per cent in the comparator group. But of the survivors, 11 could walk independently or with assistance in the screening group, compared with just one child diagnosed via a traditional clinical pathway.

“Even though they are younger, the newborn screening group were almost always more functionally able and had fewer comorbidities than those in the comparator group who were older,” says Dr Kariyawasam.

“It shows that changing the paradigm of diagnosis to being more proactive can improve the lives of children born with spinal muscular atrophy.”

The study found the need for respiratory and feeding support was also seven times less in the screening group.

“The burden of care and the morbidity of SMA was drastically lower in the newborn screening group, which has a huge effect on the quality of life for children and their families,” says Prof. Farrar.

Babies screened for SMA in NBS programs have better health outcomes than those with a later clinical diagnosis. However, forty per cent of the screening group in the study already had signs and symptoms of SMA within the first six weeks of life, showing how early the disease can start.

“Those with disease onset who get therapeutic intervention continue to gain motor skills, but we do see signs of weakness, the need for feeding and breathing support, compared to those who are treated before signs and symptoms,” says Dr Kariyawasam.

The researchers say NBS for SMA reduces clinical uncertainties that come with delayed diagnosis and help to identify the right treatment pathways.

“NBS for SMA allows us to provide more precise care and set realistic expectations about the likely health outcomes, which can help families deal with some of the uncertainty that comes with such a shocking diagnosis,” says Prof. Farrar.

Research changing rare genetic diseases

Since 2018, more than 400,000 babies have been screened for SMA in the NSW NBS program. NSW was the first jurisdiction in Australia to pilot testing for SMA in the NSW NBS program and has since committed $1.3million annually to ongoing screening. From 1 July 2022, parents of all new babies born in NSW and the ACT are offered free tests for SMA within 48-72 hours of their baby’s birth. NBS for SMA has since been recommended by the Australian Government to be rolled out nationally with support from the research team, who will help develop standardised guidelines.

However, NBS for SMA is still only available in a handful of countries worldwide. The researchers estimate fewer than 5 per cent of newborns are screened for the condition.

“This study helps to build the evidence that newborn screening is an approach that helps achieve better health outcomes for children with SMA and can encourage other countries to adopt it as a standard of care in their country,” says Prof. Farrar.

The research team are also investigating the viability of other genetic diseases to be incorporated into NBS programs.

“All the work is a multidisciplinary effort between scientists, screening clinicians and allied health, demonstrating that clinical research can make a real positive difference to the lives of children and their families,” says Dr Kariyawasam.

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Media contact:
Ben Knight
UNSW News & Content
b.knight@unsw.edu.au
(02) 9065 4915

Notes to journalists:
Dr Didu (Sandi) Kariyawasam is available for interview on request.

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