Genetic testing: SMA added to newborn heel prick in 'profound' change in medical screening

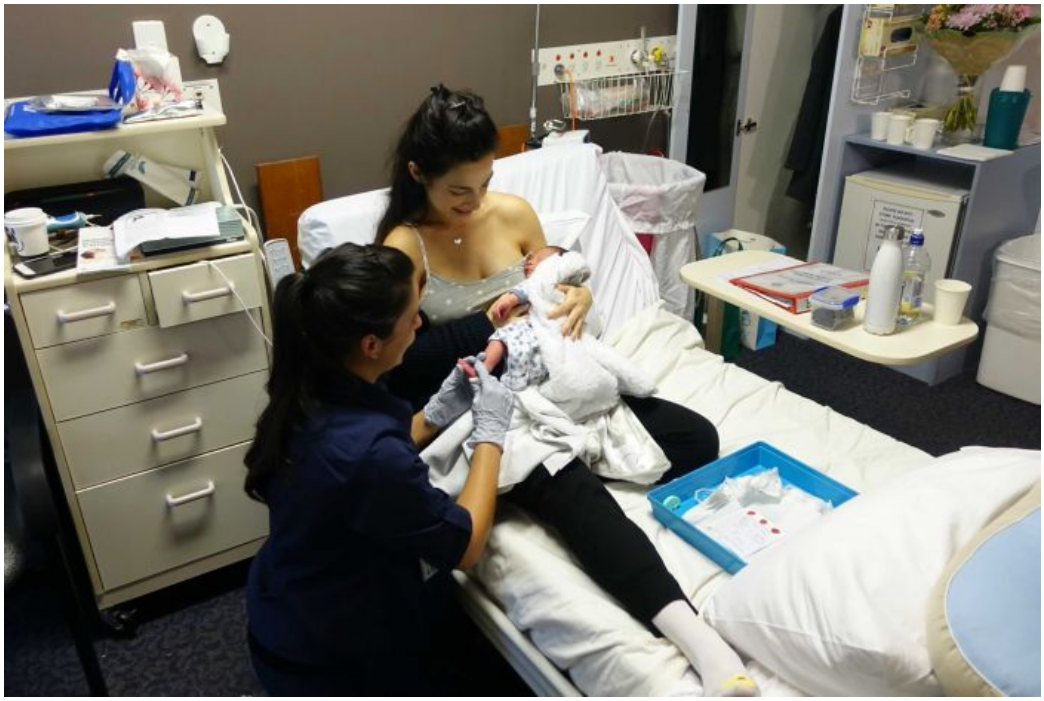

PHOTO: Lauren holds two-day-old Levendi as midwife Lindsey conducts a heel prick test to screen for a range of conditions. (ABC News: Emily Clark)

In an Australian first, newborn babies are now being routinely screened for the genetic condition SMA, or spinal muscular atrophy, and one professor says the impact on families "is going to be profound".

It is the leading genetic cause of infant death in Australia, but breakthrough treatments can now improve symptoms and slow the progression of the illness if it is diagnosed early.

For some parents, that has been a difficult and devastating process, but now, a two-year pilot program will see more than 200,000 babies born in

NSW and the ACT tested for the condition as soon as they are born, with SMA added to the newborn heel prick test.

Every newborn in Australia is offered the test, where drops of blood from the baby's foot are tested for rare but serious conditions such as PKU, hypothyroidism and cystic fibrosis.

NSW Health Minister Brad Hazzard said one baby in NSW had already been identified as having SMA since the screening was introduced, giving the newborn a chance to be treated and have a healthy life.

He urged his state and territory counterparts to "come on board with a testing program".

PHOTO: Baby Levendi receives a heel prick test to screen for a range of conditions. (ABC News: Emily Clark)

'One GP told me I was a silly first-time mum'

Haley McLean knew something was not quite right with her newborn Ruby, but doctors dismissed her concerns.

"I remember one GP told me I was a silly first-time mum," she said.

"I got told by a paediatrician to go to Bunnings, get some building materials, build a bridge and get over it."

But she persisted and after nine months, the family received the devastating diagnosis that Ruby had spinal muscular atrophy or SMA.

"Your life is just shattered into pieces and your little girl's whole life is ruined," Ms McLean said.

SMA is caused by the death of nerve cells that feed muscles, known as motor neurons.

Professor Ian Alexander, head of the Gene Therapy Research Unit at the Children's Medical Research Institute, said once the cells were lost, they were lost forever.

"The key with newborn screening is it allows you to detect the condition before the child develops symptoms, and intervene ... so that the child has lost no or minimal function," he said.

Ms McLean said she wished her daughter had access to newborn screening.

"Having this screening will save parents a lot of heartache," Ms McLean said.

Since being diagnosed, Ruby has been able to start taking a new treatment for SMA, which has helped. Dr Hooling Teoh from Sydney Children's Hospital at Randwick is her doctor.

"Now we have a treatment. We know ... the earlier you treat babies, the better the outcome," she said.

PHOTO: Ruby was diagnosed with SMA when she was nine months old. (ABC News: Rebecca Armitage)

PHOTO: A NSW newborn screening program kit includes a card with circles where drops of blood are collected and tested for a range of conditions. (ABC News: Emily Clark)

Professor Alexander said having newborn screening would make a huge difference for families.

"The impact is going to be profound. We are taking a condition with a very sad outcome and converting it to the possibility that children can live normal, healthy lives," he said.

There are also exciting new gene therapy treatments being trialled for SMA.

Forty-five patients across the world including some from Australia are being recruited for the international study. A previous small trial of gene therapy found it helped all 15 children who were treated.

"In that trial, there were infants walking and talking that would have otherwise succumbed to the disease," Professor Alexander said.

'I don't think there's anything sadder'

Mr Hazzard said he introduced the screening after meeting Rachael and Jonny Casella whose baby, MacKenzie, died from SMA at seven months.

PHOTO: Rachael and Jonny Casella lost their seven-month-old daughter Mackenzie last year. (ABC News: Sophie Scott)

"As soon as I met Mackenzie's parents, I knew we had to do something," Mr Hazzard said.

"I don't think there is anything sadder than having what appears to be a perfectly healthy baby and to find out in subsequent years that the baby you had is going to pass away from a disease that wasn't identified early enough."

In January, ABC's 7.30 program revealed the Casella's story inspired Federal Health Minister Greg Hunt to take action to help identify genetic conditions.
Dubbed "Mackenzie's Mission", the Government will spend $20 million on a pre-conception screening trial for rare and debilitating birth disorders, including SMA, Fragile X and Cystic Fibrosis.

Pre-pregnancy genetic screening can avoid babies being born with serious genetic conditions such as SMA.

At the moment, blood tests are available, but they can cost several hundred dollars and are not offered routinely.

After meeting the Casella's and hearing about the death of their daughter, Mr Hunt launched a major genetic screening program that he dubbed "Mackenzie's mission".

In the pilot study, hundreds of couples across Australia will receive pre-pregnancy genetic screening for conditions such as SMA.