childhood dementia INITIATIVE

Newborn screening and childhood dementia

Australia has a strong newborn screening program but the addition of new conditions has been slow in recent years, not keeping up with advances in treatment.

The Australian Newborn Screening Program recommends 28 conditions are screened, the majority of these have been implemented by all states and territories. This results in the detection of more than 300 babies each year who then have access to early, life saving and life changing treatments. Approximately half of these babies would otherwise suffer progressive brain damage and childhood dementia. This includes babies diagnosed with conditions such as phenylketonuria (PKU) and maple syrup urine disease.

Prior to his election in May 2022, the Australian prime minister, Anthony Albanese, pledged to expand the current newborn screening programme. An Anthony Albanese press release in April 2022 said "Australia's screening program hasn't been updated since the 1980s and, in most States and Territories, only tests for 25 conditions compared to world's best practice of screening for 80 conditions."

We compared Australian newborn screening (28 conditions) to the USA (80 conditions in some states, for example California). Careful inspection revealed discrepancies in the way that different countries and states count the number of conditions screened. For example in Australia "phenylketonuria (classical,

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intermediate and some pterin defects)" is counted as one condition but in California this is counted as four conditions

We identified 6 conditions that are widely screened in the USA that warrant urgent consideration for addition to the Australian newborn screening program:

- Sickle cell disease and beta-thalassemia
- X-linked adrenoleukodystrophy*
- Biotinidase deficiency*
- Pompe disease
- Hurler syndrome (MPS I)*
- Hunter syndrome (MPS II)*

Four of these six conditions cause childhood dementia (denoted by *). There are multiple other childhood dementia conditions that have treatments and emerging treatments requiring early diagnosis for successful treatment. This includes conditions such as CLN2 (a type of Batten disease), Niemann-Pick disease, Sanfilippo syndrome (MPS III), Krabbe, metachromatic leukodystrophy, Sly syndrome (MPS VII) and Gaucher disease. If all of these conditions were included in newborn screening, almost a quarter of the children born each year with a condition that causes childhood dementia would have treatment or clinical trial options that could significantly improve their lives.

Sickle cell disease, beta-thalassemia and X-linked adrenoleukodystrophy are already currently being considered by the Medical Services Advisory Committee (MSAC) and is tabled for the MSAC Meeting on 27-28 July 2023. This is the first time that MSAC has been used as the mechanism to consider the addition of new conditions to the newborn screening program and we eagerly await news of the decision.

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Our understanding is that the Department of Health has been working to make sure that all Australian states and territories are screening for the same conditions. They have also been assessing the infrastructure in place in each state and what is needed to implement more tests. Addressing these gaps is essential for the timely implementation of any new tests that are recommended by MSAC.

As genomic newborn screening technology advances to allow broad screening at scale, we support reconsideration of screening principles to include those without an available treatment. This gives widespread benefits to families including but not limited to avoiding long diagnostic odysseys and enabling participation in clinical trials. With appropriate processes and informed consent in place, this would be transformative for our community.

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