

Expanding Australian newborn bloodspot screening programs – making international comparisons

The Australian Government (the Government) has [delivered on its commitment to expand and achieve consistency in newborn bloodspot screening](#) (NBS) programs across Australia.

When the Government made this commitment, California screened for 80 conditions and was seen as world leading. There is no internationally agreed list of conditions for newborn screening programs, and there is considerable variation between countries. For example, the United Kingdom screens for 10 conditions, and the Netherlands screens for 27 conditions.

As of September 2026, California now includes 88 conditions in its NBS program, and all of these have been considered or are under consideration in Australia.

However, due to differences in how conditions are named and counted, direct comparison between programs is complex.

The numbers in the following tables should only be used when comparing Australian NBS to Californian NBS. See the [NBS conditions screened page](#) for Australian program counting.

Comparing Australian and California NBS programs

All conditions screened by the Californian NBS program have been considered or are under consideration for the Australian NBS programs. California currently includes 88 conditions in its NBS program.

This table compares the status of the 88 Californian NBS conditions in Australia and California.

Australian NBS Conditions	California NBS Conditions
Target: 43 Target (agreed for implementation): 4 Non-target: 28 Target under consideration: 2 Total conditions: 77 Not suitable for screening in Australia at this time: 11 Conditions screened using non-bloodspot methods: 2	Core: 36 Secondary: 50 Core agreed for implementation: 2 Total conditions: 88 Conditions screened using non-bloodspot methods: 2

Australian NBS has also included or considered additional conditions which are not in the Californian NBS program. This table shows the status of all conditions included in the Californian NBS programs, **as well as additional conditions included in Australian NBS programs.**

Australian NBS Conditions	Californian NBS Conditions
Target (screened): 48 Target (agreed for implementation): 4 Non-target: 34 Total conditions included in Australian NBS: 86 Conditions under consideration: 5 Not suitable for screening in Australia at this time: 16 Conditions screened using non-bloodspot methods: 2	Core: 36 Secondary: 50 Agreed for implementation: 2 Total conditions included in California NBS: 88 Conditions screened using non-bloodspot methods: 2

Definitions

A condition may be counted differently in each program because one program may list variants separately while another groups them under one broader condition. For this reason, the detailed tables below explain the Australian classification for each Californian condition.

Term	Definition
Target / core	The program is specifically designed to look for the condition.
Non-target / secondary	The condition may be found while screening for another condition, but it is not the main purpose of the test.
Agreed for implementation	The condition has been agreed for addition, but implementation may still be underway.
Under consideration	The condition is being assessed through Australia's decision-making pathway.
Not suitable for screening	Australia has considered the condition and determined it is not suitable for inclusion in newborn bloodspot screening at this time, based on alignment with the National Policy Framework.

Sources:

California Department of Public Health, 30 June 2026, *Recommended Uniform Screening Panel*. Available at:

<https://www.cdph.ca.gov/Programs/CFH/DGDS/Pages/nbs/Recommended-Uniform-Screening-Panel.aspx> [Accessed 03/09/2026]

Department of Health, Disability and Ageing, 26 August 2026, *What is screened in the program?* Available at:

<https://www.health.gov.au/our-work/newborn-bloodspot-screening/what-is-screened> [Accessed 03/09/2026]

Detailed Tables Index

Table	Name
Table 1	Comparing the status of California's <u>core</u> conditions to Australian NBS
Table 2	Comparing the status of California's <u>secondary</u> conditions to Australian NBS
Table 3	Comparing the status of conditions <u>under implementation</u> in California to Australian NBS
Table 4	The status of conditions <u>not included in California</u> in Australian NBS

Table 1: Comparing the status of California's core conditions to Australian NBS

Australian NBS Status / Notes	California core conditions
Target (screened): 29 conditions <i>Note on Sickle cell disease (SCD):</i> In Australia, 10 variants of SCD will be detected as target conditions. For a list of the variants please view the NBS variant fact sheet.	1. Propionic Acidemia 2. Methylmalonic Acidemia (Methylmalonyl-CoA Mutase) 3. Methylmalonic Acidemia (Cobalamin Disorders) 4. Isovaleric Acidemia 5. 3-Hydroxy-3-Methylglutaric Aciduria 6. Holocarboxylase Synthase Deficiency 7. β-Ketothiolase Deficiency 8. Glutaric Acidemia Type I 9. Carnitine Uptake Defect 10. Medium-chain Acyl-CoA Dehydrogenase Deficiency 11. Very Long-chain Acyl-CoA Dehydrogenase Deficiency 12. Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency 13. Trifunctional Protein Deficiency 14. Argininosuccinic Aciduria 15. Citrullinemia Type I 16. Maple Syrup Urine Disease 17. Homocystinuria 18. Classic Phenylketonuria 19. Tyrosinemia Type I 20. Primary Congenital Hypothyroidism 21. Congenital Adrenal Hyperplasia 22. Cystic Fibrosis - Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) 23. Classic Galactosemia 24. Guanidinoacetate methyltransferase deficiency (GAMT) 25. Severe Combined Immunodeficiencies 26. Spinal Muscular Atrophy 27. S,S Disease (Sickle Cell Anemia) 28. S, β-Thalassemia 29. S,C Disease
Target (agreed for implementation): 4 conditions	30. Biotinidase Deficiency 31. Mucopolysaccharidosis Type I 32. Mucopolysaccharidosis Type II 33. X-linked Adrenoleukodystrophy
Non-target: 1 condition	34. 3-Methylcrotonyl-CoA Carboxylase Deficiency
Not suitable for screening at this time: 2 conditions	35. Glycogen Storage Disease Type II (Pompe disease) 36. Infantile Krabbe
Screened using non-bloodspot methods: 2 conditions <i>Note on CCHD and hearing loss:</i> These conditions are screened in Australia but are not screened using bloodspot samples or counted in NBS programs.	37. Critical Congenital Heart Disease (CCHD) 38. Hearing Loss
Sub totals	
Australia	California
Target (screened): 29 Target (agreed for implementation): 4 Non-target: 1 Total conditions included in Australian NBS: 34 Conditions screened using non-bloodspot methods: 2 Not suitable for screening in Australia at this time: 2	Total California core NBS conditions: 36 Total California core conditions (including CCHD and hearing loss): 38

Table 2: Comparing the status of California's secondary conditions to Australian NBS

Australian NBS Status / Notes	California secondary conditions
Target (screened): 14 conditions <i>Note on bipterin defects and benign hyperphenylalaninaemias:</i> These conditions are included in target screening in Australia, under the same condition name of (Classic) Phenylketonuria (PKU) - including hyperphenylalaninaemias (PAH and pterin enzyme deficiencies) <i>Note on Sickle cell disease (SCD):</i> In Australia, 10 variants of SCD will be detected as target conditions. For a list of the variants please view the NBS variant fact sheet.	1. Carnitine Acylcarnitine Translocase Deficiency 2. Carnitine Palmitoyltransferase I Deficiency 3. Carnitine Palmitoyltransferase II Deficiency 4. Glutaric Acidemia Type II 5. Biotpterin Defect in Cofactor Biosynthesis 6. Biotpterin Defect in Cofactor Regeneration 7. Benign Hyperphenylalaninemia 8. Methylmalonic Acidemia with Homocystin 9. Tyrosinemia Type II 10. Tyrosinemia Type III 11. Remethylation Defects (MTHFR, MTR, MTRR, Cbl D v1, Cbl G Deficiencies) 12. Hemoglobin SD Disease 13. Hemoglobin SE Disease 14. Hemoglobin S, Variant
Non-target: 27 conditions <i>Note on Beta-thalassaemias:</i> In Australia, 8 variants of beta-thalassaemia may be incidentally detected. For a list of the variants please view the NBS variant fact sheet. <i>Note on Other haemoglobinopathies:</i> In Australia, 7 variants of other haemoglobinopathies may be incidentally detected. For a list of the variants please view the NBS variant fact sheet.	15. Malonic Acidemia 16. 2-Methyl-3-Hydroxybutyric Aciduria 17. 2-Methylbutyrylglutamic Aciduria 18. 3-Methylglutaconic Aciduria 19. Medium/Short-Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency 20. Argininemia 21. Citrullinemia Type II 22. Hypermethioninemia 23. T-Cell Related Lymphocyte Deficiencies 24. Ethylmalonic Encephalopathy 25. Alpha Thalassemia Major 26. Hemoglobin H Disease 27. Hemoglobin C Disease 28. Hemoglobin D Disease 29. Hemoglobin E, E 30. Hemoglobin Variant, Variant 31. Hereditary Persistence of Fetal Hemoglobin (HPFH)/HPFH 32. Hereditary Persistence of Fetal Hemoglobin S/HPFH 33. Hemoglobin C Beta Thalassemia 34. Hemoglobin D Beta Thalassemia 35. Hemoglobin E Beta Thalassemia 36. Hemoglobin E Delta-Beta Thalassemia 37. Hemoglobin Variant/Beta Thalassemia 38. Beta Thalassemia Major 39. Tyrosinemia, Transient 40. Congenital Adrenal Hyperplasia (11βMonooxygenase Deficiency) 41. Duarte Galactosemia
Not suitable for screening in Australia at this time: 9 conditions	42. Isobutyrylglutamic Aciduria 43. Short Chain Acyl-CoA Dehydrogenase Deficiency 44. Formiminoglutamic acidemia 45. Carbamoylphosphate Synthetase Deficiency 46. Gyrate Atrophy of the Choroid and Retina 47. Hyperornithinemia-Hyperammonemia Homocitrullinuria Syndrome 48. Hyperprolinemia Type I 49. Hyperprolinemia Type II 50. Ornithine Transcarbamylase Deficiency
Sub totals	
Australia	California
Target (screened): 14 Non-target: 27 Total conditions included in Australian NBS: 41 Not suitable for screening in Australia at this time: 9	Total California secondary NBS conditions: 50

Table 3: Comparing the status of conditions under implementation in California to Australian NBS

Australian NBS Status	California NBS: Core conditions under implementation
Under consideration: 2 conditions	1. Duchenne Muscular Dystrophy 2. Metachromatic Leukodystrophy
	Sub totals
Australia	California
Conditions under consideration: 2	Total California core conditions under implementation: 2

Table 4: The status of conditions not included in California in Australian NBS

Australian NBS Status / Notes	Condition Names
Target (screened): 5 conditions <i>Note on Sickle cell disease (SCD):</i> <i>In Australia, 10 variants of SCD will be detected as target conditions. For a list of the variants please view the NBS variant fact sheet.</i>	1. Other Galactosemias 2. Sickle cell, haemoglobin D-Punjab / Hb S,D-Punjab 3. Sickle cell, haemoglobin O-Arab / Hb S,O-Arab 4. Sickle cell, haemoglobin Lepore / Hb S,Lepore 5. Sickle cell, $\delta\beta$ thalassaemia / Hb S, $\delta\beta$ -thalassaemia
Non-target: 6 conditions <i>Note on Other haemoglobinopathies:</i> <i>In Australia, 7 variants of other haemoglobinopathies may be incidentally detected. For a list of the variants please view the NBS variant fact sheet.</i>	1. X-linked agammaglobulinemia 2. Zellweger Spectrum Disorders 3. Thyroid dyshormonogenesis 5 4. Sickle cell carrier trait 5. β -thalassaemia intermedia / Hb β -thalassaemia, β -thalassaemia 6. β thalassaemia minor / Hb β -thalassaemia
Not suitable for screening in Australia at this time: 5 conditions	1. Gaucher disease 2. Fabry Disease 3. Acid sphingomyelinase deficiency (ASMD) (Niemann-Pick disease types A and B) 4. CLN2 (neuronal ceroid lipofuscinosis 2) – Batten disease 5. Sanfilippo syndrome (Mucopolysaccharidosis type III (MPS III))
Under consideration: 3 conditions	6. Niemann pick disease type C 7. Aromatic L-amino acid decarboxylase deficiency 8. Pyridoxine-dependent epilepsy
Sub totals	
California	Australia
Conditions included in California NBS: 0	Target (screened): 5 Non-target: 6 Total conditions included in Australian NBS: 11 Conditions under consideration: 3 Not suitable for screening in Australia at this time: 5