



NBS conditions screened in Australia

September 2022

	Screened conditions	NSW/ACT	QLD/NT	SA/NT/TAS	VIC	WA
1	Argininosuccinic aciduria (ASA lyase deficiency)	yes	yes	yes	yes	yes
2	Citrullinemia (argininosuccinate synthase deficiency, citrin deficiency I and II)	yes	yes	yes	yes	yes
3	Homocystinuria (cystathionine beta-synthase deficiency)	yes	yes	yes	yes	yes
4	Maple Syrup Urine Disease (classical and variant)	yes	yes	yes	yes	yes
5	Phenylketonuria (classical, intermediate and some pterin defects)	yes	yes	yes	yes	yes
6	Congenital adrenal hyperplasia (salt wasting) (CAH)	yes	yes	yes	yes	yes
7	(Primary) congenital hypothyroidism (CH)	yes	yes	yes	yes	yes
8	Cystic fibrosis (CF)	yes	yes	yes	yes	yes
9	Carnitine/acylcarnitine translocase deficiency	yes	yes	yes	yes	yes
10	CPT-1 deficiency (carnitine palmitoyl transferase deficiency 1)	yes	yes	yes	yes	yes
11	CPT-2 deficiency (carnitine palmitoyl transferase deficiency 2)	yes	yes	yes	yes	yes
12	LCHADD (3-hydroxy long chain acyl-CoA dehydrogenase deficiency)/ TFP (trifunctional protein deficiency)	yes	yes	yes	yes	yes
13	MADD (multiple acyl-CoA dehydrogenase deficiency)/Glutaric acidemia Type 2)	yes	yes	yes	yes	yes
14	MCADD (medium chain acyl-CoA-dehydrogenase deficiency)	yes	yes	yes	yes	yes

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15	VLCADD (very long chain acyl-CoA-dehydrogenase deficiency)	yes	yes	yes	yes	yes
16**	Galactosemias	yes	yes	yes	no	yes
17	Glutaryl-CoA dehydrogenase deficiency (glutaric acidemia Type 1)	yes	yes	yes	yes	yes
18	Isovaleric acidemia	yes	yes	yes	yes	yes
19	Methylmalonic acidurias (Mutase deficiency, Cobalamin A&B deficiencies, plus Cobalamin C deficiency and related conditions)	yes	yes	yes	yes	yes
20	Propionic academia	yes	yes	yes	yes	yes
21	Holocarboxylase synthase deficiency	yes	yes	yes	yes	yes
22***	SCID	yes	planned	no	planned	planned
23	Argininemia (arginase deficiency)	yes	yes	yes	yes	yes
24	Carnitine transporter defect	yes	yes	yes	yes	yes
25	3-methylglutaconyl-CoA hydratase deficiency	yes	yes	yes	yes	yes
26#	SMA (spinal muscular atrophy)	yes	planned	no	planned	yes
27	Tyrosinemia type 1 (Fumaryl acetoacetase deficiency)	yes	yes	yes	yes	yes
28	Tyrosine aminotransferase deficiency (tyrosinemia Type 2)	yes	yes	yes	yes	yes

Notes: 1. ACT, Tas and NT do not have their own laboratories; samples are sent interstate for screening.

2. This list is accurate of 15 September 2022 and will continue to be updated should further information becomes available.

	Commonwealth has assessed condition. Jurisdiction has made a commitment to implement.
	Screening not offered.

** currently diagnosed in VIC by a medical professional

*** C'wealth assessed SCID in 2022 and found merit in screening; VIC will commence pilot screening in 2023, QLD has announced screening in 2023, WA to endorse in 2023

SMA pilot in WA. VIC will commence pilot screening in 2023. QLD has announced screening will commence in 2023.