Included conditions in neonatal screening programs

BETTER ACCESS AUSTRALIA

Australia[1]	California[2,3,4]
3-MCC (3-methylcrotonyl-CoA carboxylase deficiency)*	3-MCC (3-methylcrotonyl-CoA carboxylase deficiency)
3-methyglutaconic aciduria	3-methyglutaconic aciduria†
Argininemia (arginase deficiency)	Adrenoleukodystrophy (X-linked adrenoleukodystrophy)
Argininosuccinic aciduria (ASA lyase deficiency)	Alpha thalassemia major
Beta-ketothiolase deficiency	Argininemia (arginase deficiency)
Carnitine transporter defect	Argininosuccinic aciduria (ASA lyase deficiency)
Carnitine/acylcarnitine translocase deficiency	Benign hyperphenylalaninemia‡
Citrullinemia type I	Beta-ketothiolase deficiency
Citrullinemia type II	Biopterin defect in cofactor biosynthesis
Congenital adrenal hyperplasia*	Biopterin defect in cofactor regeneration
Congenital hypothyroidism (primary)	Biotinidase deficiency
CPT-I deficiency (carnitine palmitoyl transferase deficiency 1)	Carbamoyl phosphate synthetase deficiency
	Carnitine transporter defect
CPT-II deficiency (carnitine palmitoyl transferase deficiency 2)	Carnitine/acylcarnitine translocase deficiency
	CFTR-related metabolic syndrome
Cystic fibrosis	Citrullinemia type I
Galactosemia*	Citrullinemia type II
Glutaric acidemia type I	Congenital adrenal hyperplasia
HMGCoA lyase deficiency (Hydroxymethylglutaric aciduria)	Congenital adrenal hyperplasia (11 beta-monooxygenase deficiency)
Holocarboxylase synthetase deficiency	Congenital hypothyroidism (primary)
Homocystinuria	CPT-I deficiency (carnitine palmitoyl transferase deficiency 1)
Isobutyryl-CoA dehydrogenase deficiency	
Isovaleric acidemia	CPT-II deficiency (carnitine palmitoyl transferase deficiency 2)
LCHAD (3-hydroxy long chain acyl-CoA-dehydrogenase deficiency)	
	Cystic fibrosis
MADD (multiple acyl-CoA dehydrogenase deficiency/Glutaric acidemia type II)	Duarte galactosemia
	Ethylmalonic encephalopathy
Maple syrup urine disease	FIGLU (Formiminoglutamic acidemia)
MCADD (medium chain acyl-CoA-dehydrogenase deficiency)	Galactosemia
	Glutaric acidemia type I
Methylmalonic acidurias (Mutase deficiency, Cobalamin A&B deficiencies, plus Cobalamin C deficiency and related conditions)	Gyrate atrophy of the choroid and retina
	Hemoglobin C beta-thalassemia
	Hemoglobin C disease‡
Phenylketonuria	Hemoglobin D beta-thalassemia‡
Propionic acidemia	Hemoglobin D disease
SBCHAD (Short/branched chain acyl-CoA dehydrogenase deficiency)	Hemoglobin E beta-thalassemia
	Hemoglobin E beta-thalassemia
SCAD (Short chain acyl-CoA dehydrogenase deficiency)	Hemoglobin E delta-beta-thalassemia
SCHAD (Short chain hydroxy acyl-CoA-dehydrogenase deficiency)	Hemoglobin E, E
	Hemoglobin S, variant
SCID (Severe combined immunodeficiency)*	Hemoglobin SD disease
Spinal muscular atrophy*	Hemoglobin SE disease
Trifunctional protein deficiency	Hemoglobin variant, variant
Tyrosine aminotransferase deficiency	Hemoglobin variant/beta-thalassemia
Tyrosinemia type I	HMGCoA lyase deficiency (Hydroxymethylglutaric aciduria)
Tyrosinemia type II	Holocarboxylase synthetase deficiency
VLCADD (very long chain acyl-CoA-dehydrogenase deficiency)	Homocystinuria
	HPFH/HPFH
	Hurler syndrome (Mucopolysaccharidosis type I (MPS I))
	Hypermethioninemia
	Hyperprolinemia type I
	Hyperprolinemia type II
	Isobutyryl-CoA dehydrogenase deficiency
	Isobutyrylglycinuria
	Isovaleric acidemia
	LCHAD (3-hydroxy long chain acyl-CoA-dehydrogenase deficiency)
	M/SCHAD (Medium/short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency)

[1]HGSA, Recommendations for screening for specific disorders (August 2019)
[2]CDPH, Core conditions (October 2020)
[3]CDPH, Secondary conditions (September 2018)
[4]CDPH, Additional secondary conditions (September 2018)

	MADD (multiple acyl-CoA dehydrogenase deficiency/ Glutaric acidemia type II) Malonic acidemia Maple syrup urine disease MCADD (Medium-chain acyl-CoA-dehydrogenase deficiency) Methylmalonic acidemia Methylmalonic acidemia (Cobalamin disorders) Methylmalonic acidemia (Methylmalonyl-CoA mutase) MHBD deficiency (2-methyl-3-hydroxybutyric aciduria) Ornithine transcarbamylase deficiency Phenylketonuria Pompe (Glycogen storage disease type II) Propionic acidemia Remethylation defects (MTHFR, MTR, MTRR, Cbl D v1, Cbl G deficiencies) S/HPFH SBCHAD (Short/branched chain acyl-CoA dehydrogenase deficiency) SCAD (Short chain acyl-CoA dehydrogenase deficiency) SCID (Severe combined immunodeficiency) Sickle cell anaemia (S, S disease) Sickle cell-beta thalassemia (S, beta-thalassemia) Sickle cell-heamoglobin C disease (S, C disease) Spinal muscular atrophy T-cell related lymphocyte deficiencies Trifunctional protein deficiency Triple H syndrome (Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (HHH)) Tyrosinemia type I Tyrosinemia type II Tyrosinemia type III Tyrosinemia, transient VLCADD (very long chain acyl-CoA-dehydrogenase deficiency)

*Condition not screened for in every state/territory.
‡ No specific treatments but manage conditions through regular cardiologist visits.
‡ Does not usually require treatment.
NB: Those highlighted purple represent the four conditions without available treatment.