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Policy

Title

Counting Conditions and Summary of Conditions Screened by Programme

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Counting Conditions on Newborn Screening Panels

The way conditions are named and counted on newborn screening panels varies between programmes both nationally (eg USA, Australia) and internationally, leading to concern that infants in some jurisdictions receive 'better' screening than those in others when in fact they may be the same but listed and counted differently. For example, some counts include newborn screening other than bloodspots eg hearing and some counts include severity variations of the same disorder.

As it has been politically used as a comparison, the HGSA newborn screening committee has worked with the list from the California programme (https://www.cdph.ca.gov/Programs/CFH/DGDS/Pages/nbs/NBS-Disorders-Detectable.aspx on 01/06/2022) as it has been used as a comparator with local programmes, and agreed to list screened conditions in three categories, excluding screening which is not bloodspots although these screens (eg newborn hearing) are included in the California list. This document replaces the previous recommended disorders policy.

Category 1 Target disorders meet the following criteria

- Meet screening criteria
- There is an intent to detect with maximum sensitivity and specificity
- It is possible to determine sensitivity (which may not be high if appropriately balanced with specificity)

Disorders have been added to the target list when they have been formally approved for screening in Australia, although not all programmes may have added the disorder. Unlike lists from some other jurisdictions this list does not separate conditions with different severity with the same cause eg classical PKU would not be counted separately from hyperphenylalaninemia or salt-wasting CAH from the non-classical form.

Category 2 Incidental findings are those conditions which

- Have a marker metabolite the same as for a target condition
- May or may not benefit from early detection (may be a biochemical phenotype without a known associated clinical presentation)
- Would not meet screening criteria (eg cannot determine sensitivity)
- May or may not be detected by a programme dependent on the markers and screening algorithm for the target condition.
- May not be able to be distinguished from the target condition in the screening laboratory

Category 3 Not screened in Australasia are conditions which are screened elsewhere. The approval process is underway for some of these. The application process for adding conditions to the New Zealand panel is here https://www.nsu.govt.nz/system/files/page/newborn_metabolic_screening_programme_policy_framework_june_2011.pdf and the process and status of applications in Australia is here https://www.health.gov.au/initiatives-and-programs/newborn-bloodspot-screening/how-we-decide-what-conditions-to-test-for

Category 1 Target disorders

Inborn Errors of Metabolism

	NZ	WA	SA	QLD	NSW	VIC
Argininosuccinic Aciduria	Y	Y	Y	Y	Y	Y
Biotinidase Deficiency	Y	N	N	N	N	N
Carnitine Uptake Defect	N	Y	Y	Y	Y	Y
Carnitine Acylcarnitine Translocase Deficiency	Y	Y	Y	Y	Y	Y
Carnitine Palmitoyltransferase I Deficiency	Y	Y	Y	Y	Y	Y
Carnitine Palmitoyltransferase II Deficiency	Y	Y	Y	Y	Y	Y
Citrullinemia Type I	Y	Y	Y	Y	Y	Y
GAII ie MADD (multiple acyl-CoA-dehydrogenase deficiency)	Y	Y	Y	Y	Y	Y
(Classic) Galactosemia	Y	Y	Y	Y	Y	N
Other galactosemias (epimerase, kinase, mutarotase deficiencies)	Y	Y	Y	Y	Y	N
GAMT deficiency	N	N	N	N	N	Y
Glutaric Acidemia Type I	Y	Y	Y	Y	Y	Y
Holocarboxylase Synthase Deficiency	N	Y	Y	Y	Y	Y
Homocystinuria	Y	Y	Y	Y	Y	Y
3-Hydroxy-3-Methylglutaric Aciduria	N	Y	Y	Y	Y	Y
Isovaleric Acidemia	Y	Y	Y	Y	Y	Y
β-Ketothiolase Deficiency	N	Y	Y	Y	Y	Y
Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency	Y	Y	Y	Y	Y	Y
Maple Syrup Urine Disease	Y	Y	Y	Y	Y	Y

Medium-chain Acyl-CoA Dehydrogenase Deficiency	Y	Y	Y	Y	Y	Y
3-Methylcrotonyl-CoA carboxylase deficiency	N	Y	Y	Y	Y	N
Methylmalonic Acidemia (Methylmalonyl-CoA Mutase)	Y	Y	Y	Y	Y	Y
Methylmalonic Acidemia (Cobalamin A&B disorders)	Y	Y	Y	Y	Y	Y
Methylmalonic Acidemia Cobalamin defects C,D v2	Y	Y	Y	Y	Y	Y
Remethylation Defects (MTHFR, MTR, MTRR, Cbl D v1, Cbl G Deficiencies)	N	Y	Y	N	N	Y
(Classic) Phenylketonuria - including hyperphenylalinemias (PAH and pterin enzyme deficiencies)	Y	Y	Y	Y	Y	Y
Propionic Acidemia	Y	Y	Y	Y	Y	Y
Trifunctional Protein Deficiency	Y	Y	Y	Y	Y	Y
Tyrosinemia Type I	Y	Y	Y	N	Y	N
Tyrosinemia Type II and III	N	Y	Y	Y	Y	Y
Very Long-chain Acyl-CoA Dehydrogenase Deficiency	Y	Y	Y	Y	Y	Y

Endocrine Disorders

	NZ	WA	SA	QLD	NSW	VIC
Primary Congenital Hypothyroidism	Y	Y	Y	Y	Y	Y
Congenital Adrenal Hyperplasia (21-hydroxylase deficiency)	Y	Y	Y	Y	Y	Y

Other Disorders

	NZ	WA	SA	QLD	NSW	VIC
Cystic Fibrosis	Y	Y	Y	Y	Y	Y
Severe Combined Immunodeficiencies	Y	N	N	N	Y	N
Spinal Muscular Atrophy	N	N	N	N	Y	N

Category 2 Conditions detected while testing for target conditions

	NZ	WA	SA	QLD	NSW	VIC
Congenital Adrenal Hyperplasia (11βMonooxygenase Deficiency)	N	N	N	N	N	N
Argininemia	N	N	N	N	N	N
Carbamoylphosphate Synthetase Deficiency	N	N	N	N	N	N
Citrullinemia Type II	N	N	N	N	N	N
Ethylmalonic Encephalopathy	N	N	N	N	N	N
Formiminoglutamic acidemia	N	N	N	N	N	N
Duarte Galactosemia	N	N	N	N	N	N
Gyrate Atrophy of the Choroid and Retina	N	N	N	N	N	N
Hypermethioninemia	N	N	N	N	N	N
Hyperornithinemia-Hyperammonemia Homocitrullinuria Syndrome	N	N	N	N	N	N
Benign Hyperphenylalaninemia	N	N	N	N	N	N
Hyperprolinemia Type I	N	N	N	N	N	N

Hyperprolinemia Type II	N	N	N	N	N	N
Isobutyrylglycinuria	N	N	N	N	N	N
Malonic Acidemia	N	N	Y	Y	Y	Y
Medium/Short-Chain L-3-HydroxyacylCoA Dehydrogenase Deficiency	N	N	N	N	N	N
2-Methylbutyrylglycinuria	N	N	N	N	N	N
2-Methyl-3-Hydroxybutyric Aciduria	N	N	N	N	N	N
3-Methylglutaconic Aciduria	N	N	Y	N	N	Y
Ornithine Transcarbamylase Deficiency	N	N	N	N	N	N
Short Chain Acyl-CoA Dehydrogenase Deficiency	N	N	N	N	N	N
T-Cell Related Lymphocyte Deficiencies	N	N	N	N	N	N
Tyrosinemia, Transient	N	N	N	N	N	N
Vitamin B12 deficiency	N	N	N	N	N	N
X-linked Agammaglobulinaemia	N	N	N	N	Y	N

Category 3 Conditions screened in other jurisdictions but not Australasia

X-linked Adrenoleukodystrophy
Glycogen Storage Disease Type II (Pompe)
S,S Disease (Sickle Cell Anemia) include S,B thalassemia and S,C Disease
Various Other Hemoglobinopathies
Alpha thalassemia
Beta thalassemia
Krabbe
MPSII
Fabry
Gaucher
Nieman-Pick C
DMD
HIV
Toxoplasmosis