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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 (e.g. Australia, USA) 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 e.g. 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 (e.g. 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. For some disorders, a pilot screening programme may form part of the assessment process. Unlike lists from some other jurisdictions this list does not separate conditions with different severity with the same cause e.g. 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 or no clear evidence of response to treatment)
- 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 without confirmatory follow up testing

Australasian screening labs will detect some of these disorders, with sensitivity that will vary dependent on the technology, method and cutoffs utilised when screening for target disorders.

Category 3 Disorders under active consideration which are not current target disorders within Australasia but are screened elsewhere. 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 What is screened in the program | Australian Government Department of Health and Aged Care

Category 4 Other disorders not screened in Australasia or under active consideration as a target disorder but which are screened elsewhere. This category may include disorders for which the HGSA committee considers early detection unlikely to lead to benefit.

	NZ	WA	SA	QLD	NSW	VIC
Category 1 Target Disorders						
Inborn Errors of Metabolism						
Argininosuccinic Aciduria	Y	Y	Y	Y	Y	Y
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
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
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 II and III	N	Y	Y	Y	Y	Y
Very Long-chain Acyl-CoA Dehydrogenase Deficiency	Y	Y	Y	Y	Y	Y

Guanidinoacetate methyltransferase deficiency (Note: Because of a technology change, screening will no longer be available)	N	N	N	N	N	Y
Category 1 IEM NZ Only						
Tyrosinemia Type I	Y	N	N	N	N	N
Biotinidase Deficiency	Y	N	N	N	N	N
Category 1 Endocrine Disorders						
Primary Congenital Hypothyroidism	Y	Y	Y	Y	Y	Y
Congenital Adrenal Hyperplasia (21-hydroxylase deficiency)	Y	Y	Y	Y	Y	Y
Other Disorders						
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 Incidental findings

Congenital Adrenal Hyperplasia (11βMonooxygenase Deficiency)
Citrullinemia Type II
Ethylmalonic Encephalopathy
Formiminoglutamic acidemia
Duarte Galactosemia
Hypermethioninemia
Benign Hyperphenylalaninemia
Isobutyrylglycinuria
Malonic Acidemia
Medium/Short-Chain L-3-HydroxyacylCoA Dehydrogenase Deficiency
2-Methylbutyrylglycinuria
3-Methylcrotonyl-CoA carboxylase deficiency
2-Methyl-3-Hydroxybutyric Aciduria
3-Methylglutaconyl-CoA hydratase deficiency

Short Chain Acyl-CoA Dehydrogenase Deficiency
T-Cell Related Lymphocyte Deficiencies
Tyrosinemia Type I
Tyrosinemia, Transient
Vitamin B12 deficiency
X-linked Agammaglobulinaemia

Category 3 Conditions under active consideration in Australasia

X-linked Adrenoleukodystrophy
Sickle Cell Disease

Category 4 Other conditions screened outside of Australasia

Glycogen Storage Disease Type II (Pompe)
Various Other Hemoglobinopathies
Alpha thalassemia
Beta thalassemia
Krabbe
MPS I
MPSII
Fabry
Nieman-Pick A&B
Duchennes Muscular Dystrophy
HIV
Gaucher
Toxoplasmosis
Argininemia
Carbamoylphosphate Synthetase Deficiency
Gyrate Atrophy of the Choroid and Retina
Hyperornithinemia-Hyperammonemia
Homocitrullinuria Syndrome
Hyperprolinemia Type I
Hyperprolinemia Type II
Ornithine Transcarbamylase Deficiency