Outcomes of conditions identified through open call and horizon scanning

Condition	CAPS recommendation outcome
Metachromatic leukodystrophy (MLD)	Progressed to technical advice
Aromatic L-amino acid decarboxylase (AADC) deficiency	Progressed to technical advice
Pyridoxine-dependent epilepsy (PDE)	Progressed to technical advice
Niemann-Pick disease type C (NPC)	Progressed to technical advice
Duchenne muscular dystrophy (DMD)	Progressed to technical advice
Thyroid dyshormonogenesis 5 (TDH5)	List as a non-target condition[1]. Not referred to technical advice
X-linked agammaglobulinaemia (XLA)	Remain as a non-target condition. Not referred to technical advice
Zellweger spectrum disorders (ZSDs)	List as a non-target condition[1]. Not referred to technical advice
Infantile Krabbe disease (KD)	Not progressing to technical advice
Prader-Willi syndrome (PWS)	Not progressing to technical advice
Acrodermatitis enteropathica (AE)	Not progressing to technical advice
Chronic granulomatous disease (CGD)	Not progressing to technical advice
Familial haemophagocytic lymphohistiocytosis (fHLH)	Not progressing to technical advice
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase type 2 (HSD3B2)	Not progressing to technical advice
2,4-dienoyl-CoA reductase deficiency (DE-RED)	Not progressing to technical advice
Lysinuric protein intolerance (LPI)	Not progressing to technical advice
Gaucher disease (GD)	Not progressing to technical advice
Angelman syndrome (AS)	Not progressing to technical advice
Lysosomal acid lipase deficiency (LAL-D)	Not progressing to technical advice
Cystinosis	Not progressing to technical advice
Pyridox(am)ine 5-phosphate oxidase (PNPO) deficiency	Not progressing to technical advice
Wiskott-Aldrich syndrome (WAS)	Not progressing to technical advice
Pituitary hormone deficiency, combined or isolated, type 1 (CPHD)	Not progressing to technical advice

[1] TDH5 and Zellweger spectrum disorders will be listed as a non-target conditions because they may already be detected through existing newborn bloodspot screening for congenital hypothyroidism (CH) and X-adrenoleukodystrophy respectively.

Congenital human immunodeficiency virus (HIV)	Not progressing to technical advice
Menkes disease (MD)	Not progressing to technical advice
Congenital toxoplasmosis (CT)	Not progressing to technical advice
Congenital cytomegalovirus (cCMV)	Not progressing to technical advice
Familial glucocorticoid deficiency type 2 (FGD2)	Not progressing to technical advice
Cerebrotendinous xanthomatosis (CTX)	Not progressing to technical advice
Haemolytic anaemia / glucose-6-phosphate dehydrogenase deficiency (G6PD)	Not progressing to technical advice
Fragile X syndrome (FXS)	Not progressing to technical advice
Pseudohypoaldosteronism type 1 (PHA-1)	Not progressing to technical advice
Familial hypercholesterolaemia (FH)	Not progressing to technical advice
HMG-CoA synthase-2 deficiency (HMG-CoA)	Not progressing to technical advice
Congenital isolated adrenocorticotropic hormone (ACTH) deficiency	Not progressing to technical advice
Deficiency of adenosine deaminase 2 (DADA2)	Not progressing to technical advice
Pyrophosphatase 2 deficiency (PPA2)	Not progressing to technical advice
Common variable immunodeficiency (CVID)	Not progressing to technical advice
SLC6A1 epileptic encephalopathy	Not progressing to technical advice
Congenital disorder of glycosylation, type Ia (CDG1a)	Not progressing to technical advice
Hereditary haemochromatosis (HH)	Not progressing to technical advice
Wilson disease	Not progressing to technical advice

Outcome Summary

- 5 conditions progressing to technical advice
- 2 conditions listed as non-target conditions without referral to technical advice
- 34 conditions not progressing to technical advice
- 42 conditions in total