VCGS
leaders in genetic health

Newborn screening list of conditions

No.	Condition	Synonyms
1.	3-hydroxy-3-methylglutaryl CoA lyase	HMG CoA lyase
2.	3-methylglutaconyl CoA hydratase	3-methylglutaconic aciduria type 1
3.	Argininosuccinic aciduria	Argininosuccinate lyase
4.	Beta ketothiolase	T2 deficiency, 3-oxothiolase
5.	Carnitine palmitoyl transferase 1	CPT1
6.	Carnitine palmitoyl transferase 2	CPT2
7.	Carnitine uptake defect	CUD, systemic carnitine deficiency, carnitine transporter defect, OCTN2 defect
8.	Carnitine-acyl carnitine translocase	CACT
9.	Citrullinaemia type 1	Argininosuccinate synthetase
10.	Cobalamin disorders	cblC, cblD, cblF disease
11.	Congenital adrenal hyperplasia	Classical CAH, adrenogenital syndrome
12.	Cystic fibrosis	CFTR
13.	Glutaric aciduria type 1	Glutaryl CoA dehydrogenase, GA1
14.	Guanidinoacetate methyltransferase deficiency	GAMT
15.	Holocarboxylase synthase	HCS, multiple carboxylase deficiency, MCD
16.	Homocystinuria	Cystathionine betasynthase, CBS (vitamin responsive forms may not be detected)
17.	Isovaleryl CoA dehydrogenase	Isovaleric acidaemia, IVA
18.	Maple syrup urine disease	MSUD, branched chain keto acid dehydrogenase (mild/intermittent forms may not be detected)
19.	Medium-chain acyl CoA dehydrogenase	MCAD
20.	Methylmalonic acidaemia	Methylmalonyl CoA mutase, MMA, cblA, cblB disease
21.	Mitochodrial trifunctional protein	Long-chain hydroxy acyl carnitine dehydrogenase, LCHAD, MTP
22.	Multiple acyl CoA dehydrogenase	MADD, glutaric aciduria type 2, GA2, ETF deficiency
23.	Phenylketonuria	PKU, phenylalanine hydroxylase, including tetrahydrobiopterin defects
24.	Primary congenital hypothyroidism	CH
25.	Propionic acidaemia	Propionyl CoA carboxylase, PA, ketotic hyperglycinaemia
26.	Tyrosinaemia 2	Tyrosine aminotransferase
27.	Very long chain acyl CoA dehydrogenase	VLCAD
28.	Severe combined immunodeficiency	SCID
29.	Spinal muscular atrophy	SMA
New conditions to be implemented in 2024		
30.	Galactosaemia	Classic galactosemia, Other galactosemias (epimerase, kinase, mutarotase deficiencies)

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