VCGS
Leaders in Genetic Health

Victorian Clinical Genetics Services
Murdoch Childrens Research Institute
The Royal Children's Hospital
Flemington Road, Parkville VIC 3052
P (03) 8341 6201 W vcgs.org.au

Newborn Screening List of Conditions

Condition	Synonyms
3-hydroxy-3-methylglutaryl CoA lyase	HMG CoA lyase
3-methylglutaryl CoA hydratase	3-methylglutaconic aciduria type 1
Argininosuccinic aciduria	Argininosuccinate lyase
Citrullinaemia type 1	Argininosuccinate synthetase
Beta ketothiolase	T2 deficiency, 3-oxothiolase
Maple syrup urine disease	MSUD, branched chain keto acid dehydrogenase (mild/intermittent forms may not be detected)
Carnitine palmitoyl transferase 1	CPT1
Carnitine palmitoyl transferase 2	CPT2
Carnitine uptake defect	CUD, systemic carnitine deficiency, carnitine transporter defect, OCTN2 defect
2 Carnitine-acyl carnitine translocase	CACT
Cobalamin disorders	cblC, cblD, cblF disease
Cystic fibrosis	CFTR
Homocystinuria	Cystathionine betasynthase, CBS (vitamin responsive forms may not be detected)
Hypothyroidism	
Glutaric aciduria type 1	Glutaryl CoA dehydrogenase, GA1
Holocarboxylase synthase	HCS, multiple carboxylase deficiency, MCD
Isovaleryl CoA dehydrogenase	Isovaleric acidaemia, IVA
Medium-chain acyl CoA dehydrogenase	MCAD
Methylmalonic acidaemia	Methylmalonyl CoA mutase, MMA, cblA, cblB disease
Mitochodrial trifunctional protein	Long-chain hydroxy acyl carnitine dehydrogenase, LCHAD, MTP
Multiple acyl CoA dehydrogenase	MADD, glutaric aciduria type 2, GA2, ETF deficiency
Phenylketonuria	PKU, phenylalanine hydroxylase, including tetrahydrobiopterin defects
Propionic acidaemia	Propionyl CoA carboxylase, PA, ketotic hyperglycinaemia
Tyrosinaemia 2	Tyrosine aminotransferase
Very long chain acyl CoA dehydrogenase	VLCAD