Newborn Screening Disorders

Year Added	ACMG Code	Core Conditions	Organic acid condition	Fatty acid oxidation disorder	Amino acid disorder	ENDO Disorder	HGB Disorder	Other
2006	PROP	Propionic acidemia	X					
2006	MUT	Methylmalonic acidemia (methylmalonyl-CoA mutase)	X					
2006	Cbl A,B	Methylmalonic acidemia (cobalamin disorders)	X					
2006	IVA	Isovaleric acidemia	X					
2006	3-MCC	3-Methylcrotonyl-CoA carboxylase deficiency	X					
2006	HMG	3-Hydroxy-3-methyglutaric aciduria	X					
2006	MCD	Holocarboxylase synthase deficiency	X					
2006	ßKT	ß-Ketothiolase deficiency	X					
2006	GA1	Glutaric acidemia type I	X					
2006	CUD	Carnitine uptake defect/carnitine transport defect		X				
2006	MCAD	Medium-chain acyl-CoA dehydrogenase deficiency		X				
2006	VLCAD	Very long-chain acyl-CoA dehydrogenase deficiency		X				
2006	LCHAD	Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency		X				
2006	TFP	Trifunctional protein deficiency		X				
2006	ASA	Argininosuccinic aciduria			X			
2006	CIT	Citrullinemia, type I			X			
2006	MSUD	Maple syrup urine disease			X			
2006	HCY	Homocystinuria			X			
1965	PKU	Classic phenylketonuria			X			
2006	TYR I	Tyrosinemia, type I			X			
1978	CH	Primary congenital hypothyroidism				X		
1995	CAH	Congenital adrenal hyperplasia				X		
1988	Hb SS	S,S disease (Sickle cell anemia)					X	
1988	Hb S/ßTh	S, βeta-thalassemia					X	
1988	Hb S/C	S,C disease					X	
2005	BIOT	Biotinidase deficiency						X
2013	CCHD	Critical congenital heart disease						X
2007	CF	Cystic fibrosis						X
1978	GALT	Classic galactosemia						X
2000	HEAR	Hearing loss						X
2012	SCID	Severe combined immunodeficiency						X
2018	X-ALD	X-Linked Adrenoleukodystrophy						X
2020	MPS I	Mucopolysaccharidosis Type I						X
2020	POMPE	Pompe Disease						X
2020	SMA	Spinal Muscular Atrophy						X

Newborn Screening Disorders

Year Added	ACMG Code	Secondary Conditions	Organic acid condition	Fatty acid oxidation disorder	Amino acid disorder	ENDO disorder	HGB disorder	Other
2006	Cbl C,D	Methylmalonic acidemia with homocystinuria	X					
2006	IBG	Isobutyrylglycinuria	X					
2006	2MBG	2-Methylbutyrylglycinuria	X					
2006	3MGA	3-Methylglutaconic aciduria	X					
2006	2M3HBA	2-Methyl-3-hydroxybutyric aciduria	X					
2006	EE	Ethylmalonic encephalopathy	X					
2006	SCAD	Short-chain acyl-CoA dehydrogenase deficiency		X				
2006	GA2	Glutaric acidemia type II		X				
2006	CPT IA	Carnitine palmitoyltransferase type I deficiency		X				
2006	CPT II	Carnitine palmitoyltransferase type II deficiency		X				
2006	CACT	Carnitine acylcarnitine translocase deficiency		X				
2006	CIT II	Citrullinemia, type II			X			
2006	MET	Hypermethioninemia			X			
2006	H-PHE	Benign hyperphenylalaninemia			X			
2006	BIOPT (BS)	Biopterin defect in cofactor biosynthesis			X			
2006	BIOPT (REG)	Biopterin defect in cofactor regeneration			X			
2006	OTC	Ornitine transcarbamylase def			X			
2006	CPS	Carbamoyl phosphate synthase def			X			
2006	TYR II	Tyrosinemia, type II			X			
2006	TYR III	Tyrosinemia, type III			X			
2010	Var Hb	Various other hemoglobinopathies					X	
2012		T-cell related lymphocyte deficiencies						X

NOT on the Recommended Uniform Screening Panel

Florida Newborn Screening Program screens for 35 core conditions and 22 secondary conditions (a total of 57 conditions), 54 of which are included in the Recommended Uniform Screening Panel that is recommended by the US Department of Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children.