Supplemental Data Table S1. Disorders related to current newborn screening in Korea

Disease	OMIM No.	Gene
Amino acid disorders		
Argininosuccinic aciduria	207900	*ASL*
Citrullinemia type 1	603470	*ASS1*
Citrullinemia type 2	603471	*SLC25A13*
HHH syndrome	238970	*SLC25A15*
Homocystinuria		
Homocystinuria, B6-responsive and nonresponsive types	236200	*CBS*
Homocystinuria-megaloblastic anemia, cbl E type	236270	*MTRR*
Methylcobalamin deficiency, cblG type	250940	*MTR*
Homocystinuria due to methylenetetrahydrofolate reductase deficiency	236250	*MTHFR*
Hyperalaninemia (Pyruvate carboxylase deficiency)	266150	*PC*
Hyperargininemia	207800	*ARG1*
Hypermethioninemia		
Methionine adenosyltransferase deficiency, autosomal recessive/Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency	250850	*MAT1A*
Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	613752	*AHCY*
Hypermethioninemia due to adenosine kinase deficiency	614300	*ADK*
Hypermethioninemia with deficiency of glycine N-methyltransferase	606664	*GNMT*
Hyperornithinemia with gyrate atrophy	258870	*OAT*
Hyperprolinemia I	239500	*PRODH*
Hyperprolinemia II	239510	*ALDH4A1*
Lysinuric protein intolerance	222700	*SLC7A7*
MSUD I, II, III	248600	*BCKDHA*
		BCKDHB
		DBT
		DLD
Nonketotic hyperglycinemia	605899	*AMT*
		GLDC
		GCSH
Ornithine carbamoyltransferase deficiency	311250	*OTC*
Phenylketonuria	261600	*PAH*
Pterin disorders		
GTPCH deficiency	233910	*GCH1*
PTPS deficiency	261640	*PTS*
DHPR deficiency	261630	*QDPR*
PCD deficiency	264070	*PCBD1*
Tyrosinemia		
Tyrosinemia, type 1	276700	*FAH*
Tyrosinemia, type 2	276600	*TAT*
Tyrosinemia, type 3	276710	*HPD*
Hyperlysinemia	238700, 268700	*AASS*
D-glyceric aciduria	220120	*GLYCTK*

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Supplemental Data Table S1. Continued

Disease	OMIM No.	Gene
Histidinemia	235800	*HAL*
Pyruvate dehydrogenase E1 alpha deficiency	312170	*PDHA1*
Pyruvate dehydrogenase phosphatase deficiency	608782	*PDP1*
Pyruvate dehydrogenase E3 binding protein deficiency	245349	*PDX1*
Pyruvate dehydrogenase E2 deficiency	245348	*DLAT*
Pyruvate dehydrogenase E1 beta deficiency	614111	*PDHB*
Dicarboxylic aminoaciduria	222730	*SLC1A1*
Organic acid disorders		
2-Methylbutyryl-coenzyme A dehydrogenase deficiency	610006	*ACADSB*
3-Hydroxy-3-methylglutaryl-coenzyme A lyase deficiency	246450	*HMGCL*
3-methylcrotonyl-coenzyme A carboxylase deficiency type I	210200	*MCCC1*
3-methylcrotonyl-coenzyme A carboxylase deficiency type II	210210	*MCCC2*
3-methylglutaconyl-coenzyme A hydratase deficiency	250950	*AUH*
3-oxothiolase deficiency	203750	*ACAT1*
Biotinidase deficiency	253260	*BTD*
Glutaric acidemia type I	231670	*GCDH*
Holocarboxylase synthetase deficiency	253270	*HLCS*
Isobutyryl-CoA dehydrogenase deficiency	611283	*ACAD8*
Isovaleric Acidemia	243500	*IVD*
Methylmalonic Acidemia		
Methylmalonic aciduria, mut type	251000	*MUT*
Methylmalonic aciduria and homocystinuria, cblC type	277400	*MMACHC*
Methylmalonic aciduria, vitamin B12-responsive	251100	*MMAA*
Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	251110	*MMAB*
Methylmalonic aciduria and homocystinuria, cblD type	277410	*MMADHC*
Methylmalonic aciduria and homocystinuria, cblF type	277380	*LMBRD1*
Methylmalonyl-CoA epimerase deficiency	251120	*MCEE*
Propionic Acidemia	606054	*PCCA*
		PCCB
Fatty acid oxidation/Mitochondrial disorders		
Carnitine deficiency, systemic primary	212140	*SLC22A5*
2,4-Dienoyl-CoA reductase deficiency	222745	*DECR1*
Carnitine acylcarnitine translocase (CACT) deficiency	212138	*SLC25A20*
Carnitine palmitoyl transferase deficiency-type I deficiency	255120	*CPT1A*
Carnitine palmitoyl transferase deficiency-type II deficiency	255110, 608836	*CPT2*
MCAD deficiency	201450	*ACADM*
Short-chain acyl-coenzyme A dehydrogenase deficiency (SCAD)	201470	*ACADS*
Short-chain hydroxyacyl-coenzyme A dehydrogenase deficiency (SCHAD)	231530	*HADH*
Trifunctional protein deficiency/LCHAD deficiency	609015, 609016	*HADHA*
		HADHB

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Supplemental Data Table S1. Continued

Disease	OMIM No.	Gene
Very long-chain acyl-coenzyme A dehydrogenase deficiency (VLCAD)	201475	*ACADVL*
Ethylmalonic encephalopathy	602473	*ETHE1*
Glutaric acidemia type II	231680	*ETFDH*
		ETFA
		ETFB
Other disorders		
Galactosemia		
Galactosemia	230400	*GALT*
Galactose epimerase deficiency	230350	*GALE*
Galactokinase deficiency with cataracts	230200	*GALK1*
CAH due to 21-hydroxylase deficiency	201910	*CYP21A2*
Congenital hypothyroidism		
Hypothyroidism, congenital, nongoitrous, 1	275200	*TSHR*
Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	218700	*PAX8*
Thyroid dyshormonogenesis 1	274400	*SLC5A5*
Thyroid dyshormonogenesis 5	274900	*DUOXA2*
Thryoid dyshormonogenesis 6	607200	*DUOX2*
Thyroid dyshormonogenesis 2A	274500	*TPO*
Additional conditions		
Carbamylphosphate synthetase I deficiency	237000, 265380	*CPS1*
Malonic aciduria		*MLYCD*
Long-chain acyl-CoA dehydrogenase deficiency	609576	*ACADL*
HMG-CoA synthase deficiency	600234	*HMGCS2*
Creatine deficiency	300352	*SLC6A8*
Cystinuria	220100	*SLC3A1*
		SLC7A9
Dicarboxylic aminoaciduria	222730	*SLC1A1*
ADA deficiency	102700	*ADA*
N-acetylglutamate synthase deficiency	237310	*NAGS*