Table S1 supplementary: Genes included in PANEL 1

Genes related to NBS conditions						
Disorder type	DISEASE	Gene	MIM	Location	Gene ID	GenBank
Aminoacid metabolism	GTP CYCLOHYDROLASE I DEFICIENCY	GCH1	600225	14q22.1-q22.2	2643	NM_000161.2
Aminoacid metabolism	HOMOCYSTINURIA DUE TO CYSTATHIONINE BETA-SYNTHASE DEFICIENCY	CBS	613381	21q22.3	875	NM_001178008.1
Aminoacid metabolism	HOMOCYSTINURIA, METHIONINE SYNTHASE DEFICIENCY (cblG COMPLEMENTATION TYPE)	MTR	156570	1q43	4548	NM_000254.2
Aminoacid metabolism	HOMOCYSTINURIA, METHIONINE SYNTHASE REDUCTASE DEFICIENCY (cblE COMPLEMENTATION TYPE)	MTRR	602568	5p15.3-p15.2	4552	NM_002454.2
Aminoacid metabolism	HYPERHOMOCYSTINEMIA DUE TO DEFICIENCY OF N (5,10)-METHYLENE-TETRAHYDROFOLATE REDUCTASE ACTIVITY	MTHFR	607093	1p36.3	4524	NM_005957.4
Aminoacid metabolism	HYPERPHENYLALANINEMIA, 6-@PYRUVOYLTETRAHYDROPTERIN SYNTHASE DEFICIENCY	PTS	612719	11q22.3-q23.3	5805	NM_000317.2
Aminoacid metabolism	HYPERPHENYLALANINEMIA, DIHYDROPTERIDINE REDUCTASE DEFICIENCY	QDPR	612676	4p15.31	5860	NM_000320.2
Aminoacid metabolism	HYPERPHENYLALANINEMIA, SEPIAPTERIN REDUCTASE DEFICIENCY	SPR	182125	2p14-p12	6697	NM_003124.4
Aminoacid metabolism	HYPERPHENYLALANINEMIA: PTERIN-4-ALPHA-CARBINOLAMINE DEHYDRATASE DEFICIENCY	PCBD1	126090	10q22	5092	NM_000281.2
Aminoacid metabolism	MAPLE SYRUP URINE DISEASE (MSUD), DIHYDROLIPOAMIDE BRANCHED-CHAIN TRANSACYLASE	DBT	248610	1p31	1629	NM_001918.3
Aminoacid metabolism	MAPLE SYRUP URINE DISEASE, BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1 ALPHA POLYPEPTIDE DEFICIENCY	BCKDHA	608348	19q13.1-q13.2	593	NM_000709.2
Aminoacid metabolism	MAPLE SYRUP URINE DISEASE, BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, BETA POLYPEPTIDE (MSUD),	BCKDHB	248611	6q13-q15	594	NM_000056.2
Aminoacid metabolism	MITOCHONDRIAL PHOSPHATASE PP2Cm DEFICIENCY	PPM1K	611065	4q22.1	152926	NM_152542.4
Aminoacid metabolism	NON KETOTIC HYPERGLYCINEMIA, AMINOMETHYLTRANSFERASE DEFICIENCY (T PROTEIN)	AMT	238310	3p21.2-p21.1	275	NM_000481.2
Aminoacid metabolism	NON KETOTIC HYPERGLYCINEMIA, GLYCINE DECARBOXILASE DEFICIENCY (P PROTEIN)	GLDC	238300	9p22	2731	NM_000170.2
Aminoacid metabolism	PHENYLKETONURIA, PHENYLALANINE HYDROXYLASE DEFICIENCY	PAH	261600	12q22-q24.2	5053	NM_000277.1
Aminoacid metabolism	TYROSINEMIA TYPE I, FUMARYLACETOACETATE DEFICIENCY	FAH	276700	15q23-q25	2184	NM_000137.2
Aminoacid metabolism	TYROSINEMIA TYPE II, TYROSINE AMINOTRANSFERASE DEFICIENCY	TAT	613018	16q22.1	6898	NM_000353.2
Aminoacid metabolism	TYROSINEMIA TYPE III, 4-HYDROXYPHENYLPYRUVATE DIOXYGENASE DEFICIENCY	HPD	609695	12q24.31	3242	NM_002150.2
Energy metabolism	DEFICIENCY IN DIHYDROLIPOAMIDE DEHYDROGENASE	DLD	238331	7q31-q32	1738	NM_000108.3
Fatty Acid Metabolism	2-METHYLBUTYRYLGLYCINURIA, 2-METHYLBUTYRYL-CoA DEHYDROGENASE DEFICIENCY	ACADSB	600301	10q26.13	36	NM_001609.3

Fatty Acid Metabolism	ACYL-CoA DEHYDROGENASE MEDIUM CHAIN DEFICIENCY	**ACADM**	607008	1p31	34	NM_000016.5
Fatty Acid Metabolism	ACYL-CoA DEHYDROGENASE SHORT CHAIN DEFICIENCY	**ACADS**	606885	12q22-qter	35	NM_000017.2
Fatty Acid Metabolism	ACYL-CoA DEHYDROGENASE VERY LONG CHAIN DEFICIENCY	**ACADVL**	609575	17p13.1	37	NM_000018.2
Fatty Acid Metabolism	CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY	**CPT1A**	600528	11q13.1-q13.2	1374	NM_001876.3
Fatty Acid Metabolism	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY	**CPT2**	600650	1p32	1376	NM_000098.2
Fatty Acid Metabolism	CARNITINE TRANSPORTER DEFICIENCY	**SLC22A5**	603377	5q21	6584	NM_003060.3
Fatty Acid Metabolism	CARNITINE/ACYLCARNITINE TRANSLOCASE DEFICIENCY	**SLC25A20**	613698	3p21.31	788	NM_000387.4
Fatty Acid Metabolism	ETHYLMALONIC ENCEPHALOPATHY 1: ETHE1 DEFICIENCY	**ETHE1**	23474	19q13.31	608451	NM_014297.3
Fatty Acid Metabolism	ISOBUTYRYL-CoA DEHYDROGENASE DEFICIENCY	**ACAD8**	604773	11q25	27034	NM_014384.2
Fatty Acid Metabolism	LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY (LCHAD)	**HADHA**	600890	2p23	3030	NM_000182.4
Fatty Acid Metabolism	MULTIPLE ACYL CoA DEHYDROGENASE (MADD): ELECTRON TRANSFER FLAVOPROTEIN DEHYDROGENASE (ETFDH) DEFICIENCY. Glutaric acidemia Type II	**ETFDH**	231675	4q32-q35	2110	NM_004453.2
Fatty Acid Metabolism	MULTIPLE ACYL CoA DEHYDROGENASE (MADD): ELECTRON TRANSFER FLAVOPROTEIN, ALPHA POLYPEPTIDE(ETFA) DEFICIENCY. Glutaric acidemia Type II	**ETFA**	608053	15q23-q25	2108	NM_000126.3
Fatty Acid Metabolism	MULTIPLE ACYL CoA DEHYDROGENASE (MADD): ELECTRON TRANSFER FLAVOPROTEIN, BETA POLYPEPTIDE(ETFB) DEFICIENCY. Glutaric acidemia Type II	**ETFB**	130410	19q13.3	2109	NM_001985.2
Fatty Acid Metabolism	TRIFUNCTIONAL PROTEIN DEFICIENCY (LCHAD)	**HADHB**	143450	2p23	3032	NM_000183.2
Organic Acid Metabolism	3-METHYLCROTONYL-CoA CARBOXYLASE 1 DEFICIENCY	**MCCC1**	609010	3q27	56922	NM_020166.3
Organic Acid Metabolism	3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY	**MCCC2**	609014	5q12-q13	64087	NM_022132.3
Organic Acid Metabolism	HOLOCARBOXYLASE SYNTHETASE DEFICIENCY	**HLCS**	609018	21q22.13	3141	NM_000411.6
Organic Acid Metabolism	METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA EPIMERASE DEFICIENCY	**MCEE**	608419	2p13.3	84693	NM_032601.3
Organic Acid Metabolism	3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE deficiency	**HMGCL**	613898	1p36.1-p35	3155	NM_000191.2
Organic Acid Metabolism	AMNIONLESS DEFICIENCY	**AMN**	605799	14q32.3	81693	NM_030943.3
Organic Acid Metabolism	BETA-KETOTHIOLASE DEFICIENCY	**ACAT1**	607809	11q22.3-q23.1	38	NM_000019.3
Organic Acid Metabolism	GLUTARYL-CoA DEHYDROGENASE DEFICIENCY	**GCDH**	608801	19p13.2	2639	NM_000159.2
Organic Acid Metabolism	ISOVALERIC ACIDEMIA, ISOVALERYL COENZYME A DEHYDROGENASE DEFICIENCY	**IVD**	607036	15q14-q15	3712	NM_002225.3
Organic Acid Metabolism	MALONYL-CoA DECARBOXYLASE DEFICIENCY	**MLYCD**	606761	16q24	23417	NM_012213.2

Organic Acid Metabolism	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblF TYPE	**LMBRD1**	612625	6q13	55788	NM_018368.3
Organic Acid Metabolism	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblJ TYPE	**ABCD4**	603214	14q24.3	5826	NM_005050.3
Organic Acid Metabolism	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, CUBILIN DEFICIENCY	**CUBN**	602997	10p12.31	8029	NM_001081.3
Organic Acid Metabolism	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, INTRINSIC FACTOR DEFICIENCY	**GIF**	609342	11q13	2694	NM_005142.2
Organic Acid Metabolism	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, MMACHC DEFICIENCY (cblC COMPLEMENTATION TYPE)	**MMACHC**	609831	1p34.1	25974	NM_015506.2
Organic Acid Metabolism	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TRANSCOBALAMIN I DEFICIENCY	**TCN1**	189905	11q11-q12	6947	NM_001062.3
Organic Acid Metabolism	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TRANSCOBALAMIN II DEFICIENCY	**TCN2**	613441	22q12.2	6948	NM_000355.2
Organic Acid Metabolism	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TRANSCOBALAMIN RECEPTOR DEFICIENCY	**CD320**	606475	19p13.3-p13.2	51293	NM_016579.3
Organic Acid Metabolism	METHYLMALONIC ACIDURIA COMBINED WITH HOMOCYSTINURIA, MMADHC DEFICIENCY (cblD COMPLEMENTATION TYPE)	**MMADHC**	611935	2q23	27249	NM_015702.1
Organic Acid Metabolism	METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA MUTASE DEFICIENCY	**MUT**	609058	6p12.3	4594	NM_000255.3
Organic Acid Metabolism	METHYLMALONIC ACIDURIA, ADENOSYLTRANSFERASE DEFICIECY (cblB COMPLEMENTATION TYPE)	**MMAB**	607568	12q24	326625	NM_052845.3
Organic Acid Metabolism	METHYLMALONIC ACIDURIA, MMAA DEFICIENCY (cblA COMPLEMENTATION TYPE)	**MMAA**	607481	4q31.22	166785	NM_172250.2
Organic Acid Metabolism	METHYLMALONIC ACIDURIA, MMADHC DEFICIENCY (cblD COMPLEMENTATION TYPE)	**MMADHC**	611935	2q23	27249	NM_015702.1
Organic Acid Metabolism	MEVALONIC ACIDURIA DUE TO MEVALONATE KINASE DEFICIENCY	**MVK**	251170	12q24	4598	NM_000431.3
Organic Acid Metabolism	MITOCHONDRIAL DNA DEPLETION WITH METHYLMALONIC ACIDURIA: SUCLA2 DEFICIENCY	**SUCLA2**	603921	13q12.2-q13.3	8803	NM_003850.2
Organic Acid Metabolism	MITOCHONDRIAL DNA DEPLETION WITH METHYLMALONIC ACIDURIA: SUCLG1 DEFICIENCY	**SUCLG1**	611224	2p11.2	8802	NM_003849.3
Organic Acid Metabolism	PROPIONIC ACIDEMIA, PROPIONYL-CoA CARBOXYLASE, SUBUNIT A DEFICIENCY	**PCCA**	232000	13q32	5095	NM_000282.3
Organic Acid Metabolism	PROPIONIC ACIDEMIA, PROPIONYL-CoA CARBOXYLASE, SUBUNIT B DEFICIENCY	**PCCB**	232050	3q21-q22	5096	NM_000532.4
Urea cycle	ARGININEMIA: ARGINASE I DEFICIENCY	**ARG1**	608313	6q23	383	NM_000045.3
Urea cycle	ARGININEMIA: ARGINASE II DEFICIENCY	**ARG2**	107830	14q24.1	384	NM_001172.3
Urea cycle	ARGININOSUCCINATE LYASE DEFICIENCY	**ASL**	608310	7cen-q11.2	435	NM_000048.3
Urea cycle	CARBAMOYL PHOSPHATE SYNTHETASE DEFICIENCY	**CPS1**	608307	2q35	1373	NM_001122633.1
Urea cycle	CITRULLINEMIA TYPE II: SLC25A13 TRANSPORTER DEFICIENCY	**SLC25A13**	603859	7q21.3	10165	NM_014251.2
Urea cycle	CLASSIC CITRULLINEMIA: ARGININOSUCCINATE SHYNTASE 1 DEFICIENCY	**ASS1**	603470	9q34.1	445	NM_000050.4

Urea cycle	ORNITHINE TRANSCARBAMYLASE DEFICIENCY	OTC	300461	Xp21.1	5009	NM_000531.5

Genes not related to NBS conditions

Disorder type	DISEASE	Gene	MIM	Location	Gene ID	GenBank
Aminoacid metabolism	ALKAPTONURIA: HOMOGENTISIC ACID OXIDASE DEFICIENCY	HGD	607474	3q13.33	3081	NM_000187.3
Aminoacid metabolism	BRANCHED-CHAIN KETO ACID DEHYDROGENASE KINASE DEFICIENCY	BCKDK	614901	16p11.2	10295	NM_005881.3
Aminoacid metabolism	HYPERLYSINEMIA	AASS	605113	7q31.3	10157	NM_005763.3
Aminoacid metabolism	MULTIPLE MITOCONDRIAL DYSFUNCTIONS: SYNDROME 1 (MMDS1)	NFU1	608100	2p13.3	27247	NM_015700.2
Carbohidrate Metabolism	PYRUVATE CARBOXYLASE DEFICIENCY	PC	608786	11q13.4-q13.5	5091	NM_000920.3
Carbohidrate Metabolism	FRUCTOSEMIA, ALDOLASE B DEFICIENCY	ALDOB	612724	9q21.3-q22.2	229	NM_000035.3
Carbohidrate Metabolism	GALACTOKINASE 1 DEFICIENCY	GALK1	604313	17q24	2584	NM_000154.1
Carbohidrate Metabolism	GALACTOSE-1-PHOSPHATE URIDYLYLTRANSFERASE DEFICIENCY	GALT	606999	9p13	2592	NM_000155.2
Carbohidrate Metabolism	GLUT1 DEFICIENCY SYNDROME	SLC2A1 (GLUT1)	138140	1p35-p31.3	6513	NM_006516.2
Carbohidrate Metabolism	UDP-GALACTOSE-4-EPIMERASE DEFICIENCY	GALE	606953	1p36-p35	2582	NM_000403.3
Creatine deficiency	CREATINE TRANSPORTER DEFICIENCY	SLC6A8	300036	Xq28	6535	NM_005629.1
Creatine deficiency	GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY	GAMT	601240	19p13.3	2593	NM_000156.4
Creatine deficiency	L-ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY	GATM	602360	15q21.1	2628	NM_001482.2
Hyperoxaluria	HYPEROXALURIA TYPE 1, ALANINE-GLYOXYLATE AMINOTRANSFERASE DEFICIENCY	AGXT	604285	2q36-q37	189	NM_000030.2
Hyperoxaluria	HYPEROXALURIA TYPE II, GLYOXYLATE REDUCTASE/HYDROXYPYRUVATE REDUCTASE DEFICIENCY	GRHPR	604296	9q12	9380	NM_012203.1
Hyperoxaluria	HYPEROXALURIA TYPE III, 4-HYDROXY-2-OXOGLUTARATE ALDOLASE 1 DEFICIENCY	HOGA1	613597	10q24.1	112817	NM_138413.3
Neurotransmitters	AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY	DDC	107930	7p11	1644	NM_000790.3
Neurotransmitters	EPILEPSY PYRIDOXINE-DEPENDENT, ALDEHYDE DEHYDROGENASE 7A1 DEFICIENCY	ALDH7A1	107323	5q31	110695	NM_001182.4
Neurotransmitters	PYRIDOXAMINE 5'-PHOSPHATE OXIDASE DEFICIENCY	PNPO	603287	17q21.32	55163	NM_018129.3
Neurotransmitters	SEGAWA SYNDROME, TYROSINE HYDROXYLASE DEFICIENCY	TH	191290	11p15.5	7054	NM_000360.3
Organic Acid Metabolism	D-2-HYDROXYGLUTARIC ACIDURIA	D2HGDH	609186	2q37.3	728294	NM_152783.3

Organic Acid Metabolism	D-2-HYDROXYGLUTARIC ACIDURIA TYPE II	**IDH2**	147650	15q26.1	3418	NM_002168.2
Organic Acid Metabolism	GLUTARIC ACIDURIA III, SUCCINYL-COA:GLUTARATE-COA TRANSFERASE DEFICIENCY	**C7orf10 (SUGCT)**	609187	7p14.1	79783	NM_024728.2
Organic Acid Metabolism	L-2- HYDROXYGLUTARIC ACIDURIA: L-2-HYDROXYGLUTARATE DEHYDROGENASE	**L2HGDH**	609584	14q22.1	79944	NM_024884.2
Organic Acid Metabolism	METHYLMALONIC ACIDEMIA AND HOMOCYSTEINEMIA, cblX type	**HCFC1**	300019	Xq28	3054	NM_005334.2
Purine and Pirimidine	ADENYLOSUCCINASE DEFICIENCY	**ADSL**	608222	22q13.1	158	NM_000026.1
Purine and Pirimidine	DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY	**DPYD**	612779	1p22	1806	NM_000110.3
Purine and Pirimidine	LESCH-NYHAN SYNDROME	**HPRT1**	308000	Xq26.1	3251	NM_000194.2
Pyruvate Metabolism	PYRUVATE DEHYDEOGENASE E2 DEFICIENCY	**DLAT**	608770	11q23.1	1737	NM_001931.4
Pyruvate Metabolism	PYRUVATE DEHYDROGENASE E1-ALPHA DEFICIENCY	**PDHA1**	300502	Xp22.12	5160	NM_00284.3
Pyruvate Metabolism	PYRUVATE DEHYDROGENASE E1-BETA DEFICIENCY	**PDHB**	179060	3p21.1-p14.2	5162	NM_000925.3
Pyruvate Metabolism	PYRUVATE DEHYDROGENASE PDHX	**PDHX**	608769	11p13	8050	NM_003477.2
Transporter Disorders	CYSTINURIA, SLC3A1 TRANSPORTER DEFICIENCY	**SLC3A1**	104614	2p16.3	6519	NM_000341.3
Transporter Disorders	CYSTINURIA, SLC7A9 TRANSPORTER DEFICIENCY	**SLC7A9**	604144	19q13.1	11136	NM_014270.4
Transporter Disorders	LYSINURIC PROTEIN INTOLERANCE	**SLC7A7**	603593	14q11.2	9056	NM_001126106.1
Urea cycle	HYPERAMMONEMIA DUE TO CARBONIC ANHYDRASE VA DEFICIENCY	**CA5A**	114761	16q24.3	763	NM_001739.1
Urea cycle	HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINEMIA SYNDROME	**SLC25A15**	603861	13q14	10166	NM_014252.3
Urea cycle	N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY	**NAGS**	608300	17q21.31	162417	NM_153006.2
Vitamin Disorders	THIAMINE-RESPONSIVE MEGALOBLASTIC ANEMIA SYNDROME, SOLUTE CARRIER FAMILY 19 (THIAMINE TRANSPORTER), MEMBER 2 DEFICIENCY	**SLC19A2**	603941	1q23.3	10560	NM_006996.2
Vitamin Disorders	BASAL GANGLIA DISEASE, BIOTIN-RESPONSIVE, SOLUTE CARRIER FAMILY 19 (THIAMINE TRANSPORTER), MEMBER 3 DEFICIENCY	**SLC19A3**	606152	2q37	80704	NM_025243.3
Vitamin Disorders	BROWN-VIALETTO-VAN LAERE SYNDROME 1 AND FAZIO-LONDE DISEASE SOLUTE CARRIER FAMILY 52 (RIBOFLAVIN TRANSPORTER), MEMBER 3	**SLC52A3**	613350	20p13	113278	NM_033409.3
Vitamin Disorders	BROWN-VIALETTO-VAN LAERE SYNDROME 2 SOLUTE CARRIER FAMILY 52 (RIBOFLAVIN TRANSPORTER), MEMBER 2	**SLC52A2**	607882	8q24.3	79581	NM_024531.4
Vitamin Disorders	EXERCISE INTOLERANCE, RIBOFLAVIN-RESPONSIVE, SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER, FOLATE), MEMBER 32	**SLC25A32**	610815	8q22.3	81034	NM_030780.4
Vitamin Disorders	FLAVIN ADENINE DINUCLEOTIDE SYNTHETASE DEFICIENCY	**FLAD1**	610595	1q21.3	80308	NM_025207.4
Vitamin Disorders	RIBOFLAVIN DEFICIENCY, SOLUTE CARRIER FAMILY 52 (RIBOFLAVIN TRANSPORTER), MEMBER 1	**SLC52A1**	607883	17p13.2	55065	NM_017986.3

Vitamin Disorders	RIBOFLAVIN KINASE DEFICIENCY	**RFK**	613010	9q21.31	55312	NM_018339
Vitamin Disorders	THIAMINE METABOLISM DYSFUNCTION SYNDROME 4 SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL THIAMINE PYROPHOSPHATE CARRIER), MEMBER 19 DEFICIENCY	**SLC25A19**	606521	17q25.3	60386	NM_021734.4
Vitamin Disorders	THIAMINE METABOLISM DYSFUNCTION SYNDROME 5 : THIAMINE PYROPHOSPHOKINASE DEFICIENCY (THMD5)	**TPK1**	606370	7q34-q35	27010	NM_022445.3