Gene list of the panel

Diseases	Gene
Hemochromatosis	*HJV, HFE, TFR2, SLC40A1, HAMP*
Spherocytosis	*SPTA1, ANK1, SPTB, SLC4A1*
Pyruvate kinase deficiency	*PKLR*
Glucose-6-phosphate dehydrogenase deficiency	*G6PD*
Thalassemia	*HBA1, HBB*
Neutropenia, severe congenital	*HAX1, ELANE, G6PC3, GFI1, WAS*
Chronic granulomatous disease	*CYBA, NCF1, NCF2, NCF4, CYBB*
Bernard-Soulier syndrome	*GP1BA, GP1BB, GP9*
Glanzmann thrombasthenia	*ITGB3*
Thrombocytopenia-absent radius Syndrome	*RBM8A*
Hemophiliab	*F8, F9*
Congenital factor V deficiency	*F5*
Factor X deficiency	*F10*
Factor XIII deficiency	*F13A1, F13B*
Hypofibrinogenemia,,congenital	*FGB, FGA, FGG*
Hypoprothrombinemia	*F2*
Hemophagocytic lymphohistiocytosis, familial	*PRF1, STX11, STXBP2, UNC13D*
Fanconianemia	*FANCG, FANCC, FANCA*
Paroxysmal nocturnal hemoglobinuria	*PIGT, PIGA*
Hermansky-Pudlak syndrome	*HPS3, AP3B1, DTNBP1, HPS1, HPS6, HPS5, BLOC1S6, BLOC1S3, HPS4*
Thrombotic thrombocytopenic purpura,familial	*ADAMTS13*
Diamond-Blackfan anemia	*RPL5, RPL11, RPL35A, RPS7, RPS10, RPS17, RPS19, RPS24, RPS26*
Hereditary hemorrhagic telangiectasia syndrome	*SMAD4, ENG, ACVRL1*
Thrombocythemia	*JAK2*
Thrombophilia due to protein C deficiency	*PROC*
Thrombophilia due to protein S deficiency	*PROS1*
Thrombophilia due to antithrombin III deficiency	*SERPINC1*
Shwachman-Diamond syndrome	*SBDS*
Jervell and Lange-Nielsen syndrome	*KCNQ1, KCNE1*
Barth syndrome	*TAZ*
Heterotaxy, visceral	*ZIC3*
Myotubular myopathy	*MTM1*
Opitz GBBB syndrome	*MID1*

Cardiomyopathy, hypertrophic	*MYH7, MYBPC3, TNNT2, TNNI3*
Arrhythmogenic right ventricular dysplasia	*PKP2*
Long QT syndrome	*KCNH2*
Pulmonary hypertension	*BMPR1B, BMPR2, CAV1, CBLN2, EIF2AK4, KCNA5, KCNK3, SMAD9*
Severe combined immunodeficiency	*IL2RG, NHEJ1, DCLRE1C, RAG1, RAG2, PTPRC, IL7R, JAK3, ADA*
Lymphoproliferative syndrome	*SH2D1A, XIAP*
Agammaglobulinemia	*BTK*
Ataxia-Telangiectasia	*ATM*
Immunodeficiency	*ICOS, CD3D, LIG4*
Familial mediterranean fever	*MEFV*
Immunodeficiency with hyper-IgM	*CD40LG*
Hyper-IgE recurrent infection syndrome	*STAT3*
T-cell immunodeficiency	*FOXN1*
Chediak-Higashi syndrome	*LYST*
α-1-antitrypsin deficiency	*SERPINA1*
Lactose deficiency	*LCT*
Congenital sucrose-isomaltose deficiency	*SI*
Glucose-galactose malabsorption	*SLC5A1*
Hirschsprung disease	*RET, EDNRB, EDN3*
Fructose intolerance,Hereditary	*ALDOB*
Hyperbilirubinemia,familial	*UGT1A1, ABCC2*
Alagille syndrome	*JAG1*
Townes-Brocks syndrome	*SALL1*
Pancreatitis, hereditary	*PRSS1*
Cholestasis, progressive familial intrahepatic	*ATP8B1, ABCB11, ABCB4*
Wilson disease	*ATP7B, PRNP*
Thryoid dyshormonogenesis	*DUOX2 , SLC5A5, TG, TPO*
Congenital adrenal hypoplasia	*CYP11B1, CYP21A2, CYP17A1, NR0B1*
Pseudohypoaldosteronism	*SCNN1A, SCNN1B, SCNN1G*
Rabson-Mendenhall syndrome	*INSR*
Hypothyroidism	*TSHB, TSHR, PAX8*
Johanson-Blizzard syndrome	*UBR1*
Ehlers-Danlos syndrome, kyphoscoliotic type	*PLOD1*
Pituitary hormone deficiency	*HESX1, LHX3, POU1F1, PROP1*
Chondrodysplasia	*PTH1R, SLC26A2, FGFR3*
Spondyloepiphyseal dysplasia tarda	*TRAPPC2*
Metaphyseal chondrodysplasia	*RMRP*
Progressive pseudorheumatoid	*WISP3*
Osteopetrosis	*TCIRG1, CA2, CLCN7, OSTM1*
Osteogenesis imperfecta	*P3H1, CRTAP, COL1A1, COL1A2*

Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*
Ellis-Vancreveld syndrome	*EVC2, EVC*
Hypophosphatasia	*ALPL*
Rhizomelic chondrodysplasia punctata	*PEX7, AGPS*
Tetra-Amelia syndrome	*WNT3*
Vitamin D-dependent osteopenia	*CYP27B1, VDR*
Hypophosphatemic rickets	*DMP1, ENPP1, FGF23, PHEX, CLCN5*
Spondyloepiphyseal dysplasia	*COL2A1*
Crouzon syndrome	*FGFR2, FGFR1*
Exostoses	*EXT1, EXT2*
Epilepsy,pyridoxine-dependent	*ALDH7A1*
Friedreich ataxia	*FXN*
Spasticataxia,Charlevoix-Saguenay type	*SACS*
HARP syndrome	*PANK2*
Spastic paraplegia	*SPG7, SPAST, ATL1, REEP1, ZFYVE26, SPG11, PLP1*
Parkinson disease	*PARK7, LRRK2, PINK1, SNCA, UCHL1, PARK2*
Agenesis of the corpus callosum	*L1CAM, SLC12A6*
Arts syndrome	*PRPS1*
Ataxia,early-onset,with oculomotor apraxia and hypoalbuminemia	*APTX*
Cockayne syndrome,typea	*ERCC8*
Coffin-Lowry syndrome	*RPS6KA3*
Dejerine-Sottas disease	*MPZ, PMP22, PRX, EGR2*
Lesch-Nyhan syndrome	*HPRT1*
Lissencephaly	*TUBA1A, DCX*
Meckel syndrome	*MKS1, RPGRIP1L*
Mental retardation, X-linked, FRAXE type	*AFF2*
Tuberoussclerosis	*TSC1, TSC2*
Neurofibromatosis	*NF1, NF2*
Ceroid lipofuscinosis,neuronal	*PPT1, TPP1, CLN3, CLN5*
Dysautonomia, familial	*IKBKAP*
Nijmegen breakage syndrome	*NBN*
Rett syndrome	*MECP2*
Pyruvate dehydrogenase E1-alpha deficiency	*PDHA1*
glaucoma,congenital	*CYP1B1*
Fraser syndrome	*FRAS1, GRIP1, FREM2*
Weill-Marchesani syndrome	*FBN1, ADAMTS10*
Deafness	*GJB2, GJB6, MYO15A, SLC26A4, TMIE, TMPRSS3, OTOF, CDH23, ATP2B2, STRC, USH1C, TECTA, PCDH15, TRIOBP, CLDN14, DFNB31, ESPN, MYO6, DFNB59, LHFPL5, KCNQ4, GJB3, DFNA5, WFS1, COCH, EYA4, MYO7A, ACTG1, TMC1, MYO1A, POU3F4,*

	COL4A6, AIFM1, SLC26A4, COL11A2, MYO7A, DFNB31
Alport syndrome	*COL4A5*
Treacher Collins syndrome	*TCOF1*
Marshall syndrome	*COL11A1*
Usher syndrome	*ADGRV1, USH1C, USH2A, CIB2, CDH23, PCDH15, SANS*
Waardenburg syndrome	*SNAI2, MITF*
Craniofacial-deafness-hand syndrome	*PAX3*
Otofaciocervical syndrome	*EYA1*
CHARGE syndrome	*CHD7*
Leber congenital amaurosis	*CRB1, RPE65, CEP290, GUCY2D*
Joubert syndrome	*ARL13B, CC2D2A, INPP5E, TMEM216, OFD1, AHI1, NPHP1, CEP290, TMEM67*
Retinitis pigmentosa	*RPE65, USH2A, RHO, CLRN1, RPGR, RP2*
Congenitalaniridia	*PAX6*
Microphthalmia,syndromic	*BCOR, STRA6*
Charcot-Marie-Tooth disease	*EGR2, FGD4, FIG4, GDAP1, HSPB1, BSCL2, HSPB8, KIF1B, LITAF, DNM2, GARS, GJB1, NEFL, MFN2, MTMR2, NDRG1, RAB7A, PMP22, PRX, TRPV4, SBF2, SH3TC2, YARS*
Cohen syndrome	*VPS13B*
Duchenne and Becker muscular dystrophy	*DMD*
Emery-Dreifuss muscular dystrophy	*EMD*
Limb-Girdle muscular dystrophy	*CAPN3, DYSF, SGCG, SGCA, SGCB, SGCD, CAV3, TTN, ANO5, FKTN*
Oculopharyngeal muscular dystrophy	*PABPN1*
Muscular dystrophy,congenital merosin-deficient	*LAMA2*
Congenital myotonia	*CLCN1*
Hyperkalemic periodic paralysis	*SCN4A*
Hypokalic periodic paralysis	*CACNA1S*
Myotonic dystrophy	*DMPK, CNBP*
Nemaline myopathy	*NEB*
Spinal muscular atrophy	*SMN1, SMN2, VAPB , UBA1, DYNC1H1*
Muscular dystrophy-dystroglycanopathy	*POMT1, POMT2*
Albinism	*MC1R, OCA2, TYR, TYR, OCA2, TYRP1, SLC45A2, SLC24A5*
Cerebro oculofacioskeletal syndrome	*ERCC6*
Xeroderma pigmentosum	*XPA, XPC, DDB2, ERCC4, ERCC5*
Dyskeratosis congenita	*TERT, TERC, CTC1, NHP2, NOP10, RTEL1, WRAP53, TINF2, DKC1*
Ectodermal dysplasia	*EDA, EDAR, EDARADD*
Epidermolysis bullosa	*COL7A1, LAMA3, LAMB3, LAMC2, COL17A1, KRT5, KRT14, PLEC*
Hutchinson-Gilford progeria	*LMNA*
Ichthyosis	*TGM1*
Sjogren-Larsson syndrome	*ALDH3A2*
Trichohepatoenteric syndrome	*TTC37, SKIV2L*

Trichothiodystrophy,photosensitive	*ERCC3, GTF2H5, ERCC2*
Ehlers-Danlos syndrome	*COL5A2, COL5A1*
Angioedema, hereditary	*SERPING1*
Alport syndrome	*COL4A3, COL4A4*
Bardet-Biedl syndrome	*BBS1, BBS10*
Cystinuria	*SLC3A1, SLC7A9*
Lowe syndrome	*OCRL*
Meckel syndrome	*MKS1, B9D2, TMEM216, TMEM67, CEP290, RPGRIP1L, CC2D2A, B9D1*
Nephronophthisis	*NPHP1, NPHS1*
Polycystic kidney	*PKHD1, PKD2, PKD1*
Porphyria,congenital erythropoietic	*UROS*
Nail-patella syndrome	*LMX1B*
Short-rib thoracic dysplasia 2	*IFT80*
Ciliary dyskinesia,primary	*DNAI1, DNAH5*
Complex IV deficiency	*SCO1, COX10 , COX15, COX6B1, FASTKD2*
Cysticfibrosis	*CFTR*
Respiratory distress syndrome in premature infants	*CTNS*
Surfactant metabolism dysfunction,pulmonary	*SFTPB, ABCA3*
Hyperphenylalaninemia	***PAH, PTS, GCH1, QDPR, PCBD1***
Sepiapterin reductase deficiency	***SPR***
Maple syrup urine disease	***BCKDHA, BCKDHB, DBT, DLD***
Carbamoyl phosphate synthetase I deficiency	***CPS1***
Ornithine transcarbamylase deficiency	***OTC***
Citrullinemia	***SLC25A13, ASS1***
Argininosuccinic aciduria	***ASL***
Argininemia	***ARG1***
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	***SLC25A15***
Ornithine aminotransferase deficiency	***OAT***
Tyrosinemia	***FAH, TAT, HPD***
Nonketotic hyperglycinemia	***GLDC, AMT, GCSH***
Homocystinuria	***CBS, MTHFR, MTR, MTRR***
Hypermethioninemia	***MAT1A, AHCY***
Alkaptonuria	***HGD***
Methylmalonic aciduria	***MUT, MMAA, MMAB, MMACHC, MMADHC, MCEE***
Propionic acidemia	***PCCA, PCCB***
Isovaleric acidemia	***IVD***
Glutaric aciduria	***GCDH, SUGCT***
3-Methylcrotonyl-CoA carboxylase	***MCCC1, MCCC2***

deficiency	
Holocarboxylase synthetase deficiency	*HLCS*
Biotinidase deficiency	*BTD*
HMG-CoA lyase deficiency	*HMGCL*
3-Methylglutaconic aciduria	*AUH, TAZ, OPA3, DNAJC19*
Alpha-methylacetoacetic aciduria	*ACAT1*
Malonyl-CoA decarboxylase deficiency	*MLYCD*
2-Methylbutyrylglycinuria	*ACADSB*
Isobutyryl-CoA dehydrogenase deficiency	*ACAD8*
Canavan disease	*ASPA*
Ethylmalonic encephalopathy	*ETHE1*
Hydroxyglutaric aciduria	*L2HGDH, D2HGDH, IDH2*
Succinic semialdehyde dehydrogenase deficiency	*ALDH5A1*
Mevalonic aciduria	*MVK*
HMG-CoA synthase-2 deficiency	*HMGCS2*
Primary carnitine deficiency	*SLC22A5*
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*
Carnitine palmitoyltransferase deficiency	*CPT1A, CPT2*
Short-chain acyl-CoA dehydrogenase deficiency	*ACADS*
Medium-chain acyl-CoA dehydrogenase deficiency	*ACADM*
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*
3-Hydroxyacyl-CoA dehydrogenase deficiency	*HADH*
Trifunctional protein deficiency	*HADHA, HADHB*
Other acyl-CoA dehydrogenase deficiency diseases	*ETFA, ETFB, ETFDH*
2,4-Dienoyl-CoA reductase deficiency	*DECR1*
Glycine N-methyltransferase deficiency	*GNMT*
Zellweger syndrome	*PEX12*
Adrenoleukodystrophy	*ABCD1*
Danon disease	*LAMP2*
Gangliosidosis	*GLB1, HEXB, HEXA*
Fabry disease	*GLA*
Gaucher disease	*GBA*
Niemann-Pick disease	*NPC1, NPC2, SMPD1*
Mucopolysaccharidosis	*IDUA, IDS, SGSH, NAGLU, HGSNAT, GNS, GALNS, ARSB, GUSB, HYAL1*

Mucolipidosis	*GNPTAB, GNPTG, MCOLN1*
Krabbe disease	*GALC*
Dysplasminogenemia	*PLG*
Metachromatic leukodystrophy	*ARSA, PSAP*
Galactose epimerase deficiency	*GALE, GALK1, GALT*
Pyruvate carboxylase deficiency	*PC*
Mannosidosis	*MAN2B1*
Glycogen storage disease	*G6PC, SLC37A4, GAA, AGL, GBE1, PYGM, PYGL, PFKM, PHKA2, PHKB, PHKG2, PHKA1, GYS2, GYS1, LDHA, ALDOA, ENO3, GYG1, SLC37A4, PGAM2*
Dihydrolipoamide dehydrogenase deficiency	*DLD*
Cholesteryl ester storage disease	*LIPA*
Hyperinsulinemic hypoglycemia, familial	*ABCC8*
Hydroxysteroid dehydrogenase	*HSD17B3, HSD17B4, HSD17B10*
GRACILE syndrome	*BCS1L*
Noonan syndrome	*BRAF, KRAS, NRAS, PTPN11, RAF1, SOS1*
Silver-Russell syndrome	*H19*
Smith-Lemli-Opitz syndrome	*DHCR7*
Bartter syndrome	*KCNJ1*
Laron dwarfism	*GHR*
Menkes disease	*ATP7A*
Molybdenum cofactor deficiency	*MOCS1, MOCS2*
Cerebrotendinous xanthomatosis	*CYP27A1*

Diseases and genes related to amino acid metabolism, organic acid metabolism, and fatty acid oxidation that may be detected by MS/MS screening applied in traditional NBS were in bold, carrier frequencies of the genes in bold were calculated.