PRIMARY IMMUNODEFICIENCY PANEL with 5 sub-panels:

Invitae Agammaglobulinemia Panel (36)
Invitae Autoinflammatory and Autoimmunity Syndromes Panel (156)
Invitae Neurodevelopmental Disorders Panel (241)
Invitae Primary Immunodeficiency Panel (429)
Invitae Supplemental Metabolic Newborn Screening Panel (192)

1. Invitae Agammaglobulinemia Panel (36)

Genes

BLM, BLNK, BTK, CCBE1, CD19, CD27, CD79A, CD79B, CD81, CDC42, CDCA7, DNMT3B, FAT4, FNIP1, GATA2, HELLS, ICOSLG, IGLL1, IL2RB, IRF2BP2, IRF4, KMT2A, LIG1, LRRC8A, MOGS, MYSM1, OAS1, PIK3R1, SEC61A1, SH2D1A, SLC39A7, TCF3, TOP2B, TRNT1, XIAP, ZBTB24

Disorders tested

- Agammaglobulinemia and hypogammaglobulinemia

2. Invitae Autoinflammatory and Autoimmunity Syndromes Panel (156)

Genes

ACP5, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, ANKZF1, AP3B1, ARPC1B, ASAH1, BACH2, BLOC1S6, BTK, C17orf62, CARD14, CARD8, CASP10, CASP8, CCBE1, CD27, CD3G, CD40, CD40LG, COPA, CR2, CTLA4, CYBA, CYBB, DCLRE1C, DDX58, DEF6, DKC1, DNASE1L3, DNASE2, DOCK8, DSG1, DUOX2, ELANE, FADD, FAS, FASLG, FCHO1, FOXP3, G6PC3, ICOS, IFIH1, IL10, IL10RA, IL10RB, IL1RN, IL21, IL21R, IL2RA, IL2RB, IL2RG, IL36RN, IRF2BP2, ITCH, ITGAM, ITGB2, ITK, JAK1, LIG4, LPIN2, LRBA, LYN, LYST, MAGT1, MEFV, MVK, NCF2, NCF4, NFAT5, NFKB1, NFKB2, NFKBIA, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OAS1, ORAI1, OTULIN, PEPD, PIK3CD, PIK3R1, PLCG2, PNP, POLA1, POMP, PRF1, PRKCD, PSMA3, PSMB4, PSMB8, PSMG2, PSTPIP1, RAB27A, RAC2, RAG1, RAG2, RASGRP1, RBCK1, RFX5, RFXANK, RFXAP, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF31, RTEL1, SAMHD1, SCO2, SH2D1A, SH3BP2, SI, SIAE, SKIV2L, SLC29A3, SLC37A4, SLC7A7, STAT1, STAT3, STAT4, STAT5B, STIM1, STX11, STXBP2, TBX1, TGFB1, TGFBR1, TGFBR2, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF6B, TNFSF12, TOP2B, TPP2, TREX1, TRNT1, TTC37, TTC7A, UNC13D, UNG, WAS, XIAP, ZAP70, ZNF341

Disorders Tested:

- monogenic autoinflammatory syndromes
- monogenic autoimmunity
- periodic fever syndromes

- familial cold autoinflammatory syndromes
- familial Mediterranean fever
- monogenic inflammatory bowel disease

3. Invitae Neurodevelopmental Disorders Panel (241)

Genes

ACTB, ACTG1, ADNP, ADSL, AGA, AHDC1, ALDH5A1, ALDH7A1, AMER1, ANKRD11, AP1S2, ARG1, ARID1A, ARID1B, ARSA, ARX, ASNS, ASXL1, ATP1A3, ATP7A, ATRX, AUTS2, BCAP31, BRAF, BRAT1, BRD4, BRWD3, CACNA1A, CACNA1E, CAMK2B, CASK, CBL, CC2D2A, CDK13, CDKL5, CHD2, CHD7, CHD8, CLCN4, CLN2 (TPP1), CLN3, CLN5, CLN6, CLTC, CNTNAP2, COL4A1, CREBBP, CTNNB1, CUL3, DDC, DDX3X, DEAF1, DHCR7, DNM1L, DNMT3A, DOCK6, DPF2, DYNC1H1, DYRK1A, EEF1A2, EFTUD2, EHMT1, EP300, EZH2, FGD1, FOLR1, FOXG1, FOXP1, GABBR2, GABRB3, GABRG2, GALC, GAMT, GATAD2B, GATM, GLB1, GM2A, GNAO1, GNAS, GNS, GPC3, GRIA3, GRIN1, GRIN2A, GRIN2B, HDAC8, HEXA, HEXB, HGSNAT, HIVEP2, HNRNPK, HNRNPU, HRAS, HUWE1, IDS, IDUA, IGF1R, IL1RAPL1, IQSEC2, ITPR1, KANSL1, KAT6A, KAT6B, KCNA2, KCNB1, KCNH1, KCNQ2, KCNT1, KDM5C, KDM6A, KIF1A, KMT2A, KMT2B, KMT2D, KMT2E, KRAS, L1CAM, LZTR1, MAGEL2, MAN1B1, MAP2K1, MAP2K2, MBD5, MECP2, MED12, MED13L, MEF2C, MFSD8, MID1, MTOR, NAA10, NAA15, NAGLU, NALCN, NEXMIF, NF1, NFIA, NFIX, NGLY1, NHS, NIPBL, NONO, NPC1, NR2F1, NRAS, NRXN1, NSD1, NSUN2, OCRL, OPHN1, OTC, PACS1, PACS2, PAH, PCBD1, PCDH19, PDHA1, PGAP3, PHF21A, PHF6, PHIP, PLA2G6, PMM2, POLG, PPM1D, PPP1CB, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PTEN, PTPN11, PTS, PURA, QDPR, RAD21, RAF1, RAI1, RBM10, RIT1, RPS6KA3, SATB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SETBP1, SETD5, SGSH, SHOC2, SIN3A, SLC13A5, SLC16A2, SLC2A1, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SON, SOS1, SOS2, SOX11, SPAST, SPATA5, SPTAN1, STAG1, STXBP1, SURF1, SYNGAP1, TAF1, TBCK, TBL1XR1, TCF20, TCF4, TELO2, TRAPPC9, TRRAP, TSC1, TSC2, TUBA1A, UBE3A, UNC80, USP9X, VPS13B, WDR45, WWOX, ZBTB18, ZBTB20, ZC4H2, ZDHHC9, ZEB2, ZIC2, ZMIZ1, ZMYND11

Disorders tested

- Aarskog-Scott syndrome
- Adenylosuccinate lyase deficiency
- Allan-Herndon-Dudley syndrome
- Alpha-thalassemia X-linked intellectual disability syndrome
- Alternating hemiplegia of childhood
- Angelman syndrome
- Arboleda-Tham syndrome
- Arginase deficiency
- Aromatic L-amino acid decarboxylase deficiency
- Asparagine synthetase deficiency
- Aspartylglucosaminuria
- Au-Kline syndrome
- Baraitser-Winter syndrome
- Beta-propeller protein-associated neurodegeneration
- Bohring-Opitz syndrome
- Bosch-Boonstra-Schaaf optic atrophy syndrome
- Cerebral creatine deficiency syndrome

- Cerebral folate deficiency
- CHARGE syndrome
- Christianson syndrome (Angelman-like syndrome)
- Chung-Jansen syndrome
- Coffin-Lowry syndrome
- Coffin-Siris syndrome
- Cohen syndrome
- COL4A1-related conditions
- Cornelia de Lange syndrome
- Costello syndrome
- Developmental and epileptic encephalopathy
- DOCK6-related Adams-Oliver syndrome
- Encephalopathy due to defective mitochondrial and peroxisomal fission
- GAND syndrome
- Genitopatellar syndrome and Say-Barber-Biesecker-Young-Simpson syndrome
- Gillespie syndrome
- Glass syndrome
- Glucose transporter type 1 (GLUT1) deficiency syndrome
- GM1-gangliosidosis
- GM2-gangliosidosis
- GNAS-related conditions
- Helsmoortel-van der Aa syndrome
- Heyn-Sproul-Jackson syndrome
- Infantile hypotonia with intellectual disability and characteristic facies
- Insulin-like growth factor I resistance
- Jansen de Vries syndrome
- Kabuki syndrome
- KBG syndrome
- KCNH1-related conditions
- Kleefstra syndrome
- KMT2B-related dystonia
- Koolen-de Vries syndrome
- Krabbe disease
- L1 syndrome
- Leigh syndrome due to mitochondrial complex IV deficiency
- Lowe syndrome
- MAN1B1-congenital disorder of glycosylation
- Mandibulofacial dysostosis-microcephaly syndrome
- MED12-related intellectual disability syndromes
- Menkes disease
- Metachromatic leukodystrophy
- Mowat-Wilson syndrome
- Mucopolysaccharidoses types I, II, III
- Myoclonic-atonic epilepsy
- NALCN-related congenital contractures of the limbs and face, hypotonia, and developmental delay
- Nance-Horan syndrome
- Neurodevelopmental and intellectual disability disorders

- Neuronal ceroid lipofuscinosis
- NF1-related conditions
- NFIA-related brain malformations and urinary tract defects
- NGLY1-related congenital disorder of deglycosylation
- Nicolaides-Baraitser syndrome
- Niemann-Pick disease type C
- Noonan syndrome
- O’Donnell-Luria-Rodan syndrome
- Ogden syndrome
- Opitz GBBB syndrome
- Ornithine transcarbamylase deficiency
- Osteopathia striata with cranial sclerosis
- PGAP3-congenital disorder of glycosylation
- Phenylketonuria
- Pierpont syndrome
- Pitt-Hopkins syndrome
- PLA2G6-related conditions
- PMM2-congenital disorder of glycosylation
- POLG-related conditions
- Primrose syndrome
- PTEN hamartoma tumor syndrome
- Pyridoxine-dependent epilepsy
- Pyruvate dehydrogenase E1-alpha (PDHE1α) deficiency
- RAI1-related conditions
- Raynaud-Claes syndrome
- Renpenning syndrome
- Rett syndrome
- Rubinstein-Taybi syndrome
- Schaaf-Yang syndrome
- Schinzel-Giedion midface retraction syndrome
- Schuurs-Hoeijmakers syndrome
- Simpson-Golabi-Behmel syndrome
- Smith-Kingsmore syndrome
- Smith-Lemli-Opitz syndrome
- Sotos syndrome
- Succinic semialdehyde dehydrogenase deficiency
- TARP syndrome
- Tatton-Brown-Rahman syndrome
- Tetrahydrobiopterin-deficient hyperphenylalaninemia
- Tuberous sclerosis complex
- Vulto-van Silfout-de Vries syndrome
- Weaver syndrome
- Wieacker-Wolff syndrome
- Wiedemann-Steiner syndrome
- Witteveen-Kolk syndrome
- Xia-Gibbs syndrome
- You-Hoover-Fong syndrome
- ZTTK syndrome

4. Invitae Primary Immunodeficiency Panel (429)

Genes

ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, ADGRE2, AICDA, AIRE, AK2, ALG6, ALPK1, ANGPT1, ANKZF1, AP3B1, AP3D1, ARHGEF1, ARPC1B, ASAH1, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BTK, C17orf62, C1QA, C1QB, C1QC, C1S, C2, C3, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD8, CARD9, CARMIL2, CASP10, CASP8, CBL, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFI, CFP, CHD7, CIB1, CIITA, CLCN7, CLPB, COL7A1, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYP27A1, DBR1, DCLRE1C, DDX58, DEF6, DGAT1, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DSG1, DTNBP1, DUOX2, EFL1, EIF2AK3, ELANE, EPG5, ERBIN, ERCC2, ERCC3, ERCC6L2, EXTL3, FADD, FANCA, FANCB, FANCE, FANCF, FANCI, FANCL, FAS, FASLG, FAT4, FCHO1, FERMT1, FERMT3, FNIP1, FOXI3, FOXN1, FOXP3, FPR1, G6PC, G6PC3, G6PD, GATA1, GATA2, GFI1, GINS1, GTF2E2, GTF2H5, GUCY2C, HAX1, HELLS, HMOX1, HPS1, HPS3, HPS4, HPS5, HPS6, HTRA2, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17F, IL17RA, IL17RC, IL18BP, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, IRAK4, IRF2BP2, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGAM, ITGB2, ITK, JAGN1, JAK1, JAK3, KAT6A, KDM6A, KMT2A, KMT2D, LAMTOR2, LAT, LCK, LCT, LIG1, LIG4, LIPA, LPIN2, LRBA, LRRC8A, LYN, LYST, MAD2L2, MAGT1, MALT1, MAP3K14, MCM4, MEFV, MKL1, MOGS, MPLKIP, MS4A1, MSN, MTHFD1, MVK, MYD88, MYO5B, MYSM1, NBAS, NBN, NCF2, NCF4, NCKAP1L, NCSTN, NEUROG3, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NSMCE3, OAS1, ORAI1, OSTM1, OTULIN, PARN, PAX1, PEPD, PGM3, PIK3CD, PIK3R1, PLCG2, PLVAP, PMM2, PNLIP, PNP, POLA1, POLD1, POLD2, POLE, POLE2, POLR3A, POLR3F, POMP, PRF1, PRKCD, PRKDC, PSENEN, PSMA3, PSMB4, PSMB8, PSMG2, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, REL, RELA, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF168, RNF31, RNU4ATAC, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SAR1B, SCO2, SEC61A1, SEMA3E, SERPING1, SGPL1, SH2D1A, SH3BP2, SH3KBP1, SI, SIAE, SKIV2L, SLC10A2, SLC26A3, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC51B, SLC5A1, SLC7A7, SLC9A3, SLX4, SMARCAL1, SMARCD2, SNX10, SP110, SPINK5, SPINT2, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT4, STAT5B, STIM1, STK4, STN1, STX11, STX3, STXBP2, TAOK2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCF3, TCIRG1, TCN2, TERC, TERT, TFRC, TGFB1, TGFBR1, TGFBR2, THBD, TICAM1, TIMM50, TINF2, TLR3, TLR7, TMC6, TMC8, TMEM173, TMPRSS15, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF6B, TNFRSF9, TNFSF11, TNFSF12, TONSL, TOP2B, TP63, TPP2, TRAF3, TRAF3IP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNC45A, UNC93B1, UNG, USB1, VAV1, VPS13B, VPS45, WAS, WDR1, WIPF1, WNT2B, WRAP53, XIAP, ZAP70, ZBTB24, ZCCHC8, ZNF341

Disorders tested

- severe combined immunodeficiency
- combined immunodeficiency
- combined immunodeficiency with syndromic features
- major histocompatibility complex class I and II deficiencies

- dyskeratosis congenita
- agammaglobulinemia and hypogammaglobulinemia
- hyper IgM syndrome
- hyper IgE syndrome, including Netherton syndrome
- monogenic common variable immune deficiency
- chronic mucocutaneous candidiasis
- herpes simplex encephalitis
- epidermodysplasia verruciformis
- WHIM syndrome
- Mendelian susceptibility to mycobacterial infections
- autoimmune lymphoproliferative syndrome
- familial hemophagocytic lymphohistiocytosis and related disorders
- Hermansky-Pudlak syndrome
- congenital neutropenia
- chronic granulomatous disease
- leukocyte adhesion deficiency
- pulmonary alveolar proteinosis
- immunodeficiency- centromeric instability- facial anomalies syndrome
- complement deficiencies
- monogenic congenital diarrhea
- monogenic autoinflammatory syndromes
- monogenic autoimmunity
- periodic fever syndromes
- familial cold autoinflammatory syndromes
- familial Mediterranean fever
- monogenic inflammatory bowel disease
- activated PI3K-delta syndrome

5. Invitae Supplemental Metabolic Newborn Screening Panel (192)

Genes

A4GALT, ACAD9, ALDH18A1, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ARCN1, ATP13A2, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V1A, ATP6V1E1, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, BOLA3, C1GALT1C1, CA5A, CAD, CANT1, CCDC115, CHST14, CHST3, CHST6, CHSY1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COPA, COPB2, CSGALNACT1, CTSD, CYP27A1, D2HGDH, DDOST, DHCR7, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, DSE, EOGT, EXT1, EXT2, EXTL3, FKRP, FKTN, FUK, FUT8, G6PC3, GALNT3, GANAB, GFPT1, GLRX5, GLUL, GM2A, GMPPA, GMPPB, GNE, GNPTAB, GNS, GORAB, GOSR2, GPAA1, HEXA, HEXB, HGSNAT, HMGCS2, IBA57, ISCA2, ISPD, JAGN1, KCTD7, LARGE1, LFNG, LIAS, LIPT1, LIPT2, MAGT1, MAN1B1, MFSD8, MGAT2, MOGS, MPDU1, MPI, NAGLU, NANS, NFU1, NGLY1, NPC1, NPC2, NUS1, OGT, OXCT1, PAPSS2, PGAP1, PGAP2, PGAP3, PGM3, PIGA, PIGB, PIGC, PIGG, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGT, PIGU, PIGV, PIGW, PIGY, PMM2, POFUT1, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPT1, PRKCSH, RFT1, RPN2, RXYLT1, SAR1B, SEC23A, SEC23B, SEC24D, SEC63, SGSH, SLC10A7, SLC16A1, SLC26A2, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A8, SLC6A9, SLC7A7, SLC9A7,

SRD5A3, SSR3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, TGDS, TMEM165, TMEM199, TRAK1, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC6B, TRAPPC9, TRIP11, TUSC3, VMA21, VPS13B, XYLT1, XYLT2

DISORDERS TESTED

- A4GALT-congenital disorder of glycosylation (A4GALT-CDG)
- Mitochondrial complex I deficiency, nuclear type 20 (MCD1N20)
- ALDH18A1-related conditions:
  - Delta-pyrroline-5-carboxylate synthetase (P5CS) deficiency
  - Cutis laxa (ADCL3 and ARCL3A)
  - Spastic paraplegia (SPG9A and SPG9B)
- ALG1-congenital disorder of glycosylation (ALG1-CDG, CDG-Ik)
- ALG11-congenital disorder of glycosylation (ALG11-CDG, CDG-Ip)
- ALG12-congenital disorder of glycosylation (ALG12-CDG, CDG-Ig)
- ALG13-congenital disorder of glycosylation (CDG-Is)
- Congenital myasthenic syndrome 15 (CMS15), ALG14-congenital disorder of glycosylation (ALG14-CDG)
- ALG2-congenital disorder of glycosylation (ALG2-CDG, CDG-Ii)
- ALG3-congenital disorder of glycosylation (CDG-Id)
- ALG6-congenital disorder of glycosylation (CDG-Ic)
- ALG8-congenital disorder of glycosylation (ALG8-CDG, CDG-Ih)
- ALG9-congenital disorder of glycosylation (ALG9-CDG, CDG-IL)
- Rhizomelic short stature with microcephaly, micrognathia and developmental delay; ARCN1-congenital disorder of glycosylation (ARCN1-CDG)
- Kufor-Rakeb syndrome (KRS), Spastic paraplegia (SPG78)
- ATP6AP1 deficiency, ATP6AP1-congenital disorder of glycosylation (ATP6AP1-CDG)
- ATP6AP2-related conditions:
  - X-linked intellectual disability with epilepsy (MRXE)
  - Glycosylation disorder with immunodeficiency, liver disease, psychomotor impairment and cutis laxa (GILPC)
- ATP6V0A2-congenital disorder of glycosylation (ATP6V0A2-CDG), Cutis laxa type 2A (ARCL2A)
- Epileptic encephalopathy of childhood onset (EEOC), ATP6V1A-congenital disorder of glycosylation (ATP6V1A-CDG), Cutis laxa type 2D (ARCL2D)
- ATP6V1E1-congenital disorder of glycosylation (ATP6V1E1-CDG), Cutis laxa type 2C (ARCL2C)
- Muscular dystrophy-dystroglycanopathy type A11 (MDDGA11)
- B3GALT6-related conditions:
  - Spondyloepimetaphyseal dysplasia with joint laxity type 1 (SEMDJL1)
  - Spondylodysplastic Ehlers-Danlos syndrome (spEDS)
  - B3GALT6-congenital disorder of glycosylation (B3GALT6-CDG)
- B3GAT3-related conditions:
  - Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects (JDSCD)
  - B3GAT3-congenital disorder of glycosylation (B3GAT3-CDG)
- Peters-plus syndrome, B3GLCT-congenital disorder of glycosylation (B3GLCT-CDG)

- B4GALNT1-congenital disorder of glycosylation (B4GALNT1-CDG), Hereditary spastic paraplegia (SPG26)
- B4GALT1-congenital disorder of glycosylation (B4GALT1-CDG, CDG-IId)
- Spondylodysplastic Ehlers-Danlos syndrome (EDS), B4GALT7-congenital disorder of glycosylation (B4GALT7-CDG)
- Muscular dystrophy-dystroglycanopathy type A13 (MDDGA13)
- Multiple mitochondrial dysfunctions syndrome 2 (MMDS2)
- Tn polyagglutination syndrome, Congenital disorder of glycosylation (CDG)
- Carbonic anhydrase VA deficiency (CAVA)
- CAD-related conditions:
    - Early infantile epileptic encephalopathy (EIEE)
    - Autism spectrum disorder with abnormal glycosylation
    - Congenital heart disease with neurodevelopmental disability
- Desbuquois dysplasia, CANT1-congenital disorder of glycosylation (CANT1-CDG)
- CCDC115-congenital disorder of glycosylation (CCDC115-CDG, CDG2o)
- CHST14-congenital disorder of glycosylation (CHST14-CDG) (Musculocontractural type Ehlers-Danlos syndrome)
- Spondyloepiphyseal dysplasia with congenital joint dislocations (SEDCJD), CHST3-congenital disorder of glycosylation (CHST3-CDG)
- Macular corneal dystrophy
- Temtamy preaxial brachydactyly syndrome (TPBS), CHSY1-congenital disorders of glycosylation (CHSY1-CDG)
- Neuronal ceroid lipofuscinosis type 3 (CLN3)
- Neuronal ceroid lipofuscinosis type 5 (CLN5)
- Neuronal ceroid lipofuscinosis type 6 (CLN6)
- Neuronal ceroid lipofuscinosis type 8 (CLN8)
- COG1-congenital disorder of glycosylation (COG1-CDG, CDG-IIg)
- COG2-congenital disorder of glycosylation (COG2-CDG, CDG-IIq)
- COG4-congenital disorder of glycosylation (CDG-IIj), Saul-Wilson syndrome (SWILS)
- COG5-congenital disorder of glycosylation (COG5-CDG, CDG-IIi)
- COG6-congenital disorder of glycosylation (COG6-CDG, CDG-IIL)
- COG7-congenital disorder of glycosylation (CDG-IIe)
- COG8-congenital disorder of glycosylation (COG8-CDG, CDG-IIh)
- Autoimmune interstitial lung, joint, and kidney disease (AILJK), COPA-congenital disorders of glycosylation (COPA-CDG)
- Congenital disorders of glycosylation (COPB2-CDG)
- Muscular dystrophy-dystroglycanopathy type A7 (MDDGA7) and C7 (MDDGC7)
- CSGALNACT1-related skeletal dysplasia, Congenital disorders of glycosylation (CSGALNACT1-CDG)
- Neuronal ceroid lipofuscinosis type 10 (CLN10)
- Cerebrotendinous xanthomatosis
- D-2-hydroxyglutaric aciduria
- DDOST-congenital disorder of glycosylation (CDG-Ir)
- Smith-Lemli-Opitz syndrome (SLOS)
- DHDDS-related conditions:
    - Retinitis pigmentosa (RP)
    - Developmental and epileptic encephalopathy syndrome
    - Congenital disorder of glycosylation (CDG-Ibb)

- DOLK-congenital disorder of glycosylation (CDG-1m)
- Congenital myasthenic syndrome 13 (CMS13), DPAGT1-congenital disorder of glycosylation (CDG-Ij)
- DPM1-congenital disorder of glycosylation (DPM1-CDG, CDG-Ie)
- DPM2-congenital disorder of glycosylation (DPM2-CDG, CDG-Iu)
- DPM3-congenital disorder of glycosylation (DPM1-CDG, CDG-Io)
- Ehlers-Danlos syndrome, musculocontractural type 2 (EDS-MC2), Congenital disorders of glycosylation (DSE-CDG)
- Adams-Oliver syndrome (AOS), Congenital disorders of glycosylation (EOGT-CDG)
- Hereditary multiple osteochondromas (HMO) (Hereditary multiple exostosis), Congenital disorder of glycosylation (EXT1-CDG and EXT2-CDG)
- EXTL3 deficiency
- Muscular dystrophy-dystroglycanopathy type A5 (MDDGA5), type B5 (MDDGB5) and type C5 (MDDGC5)
- Muscular dystrophy-dystroglycanopathy type A4 (Fukuyama congenital muscular dystrophy), type B4, and type C4
- Congenital disorder of glycosylation with defective fucosylation
- FUT8-congenital disorder of glycosylation (FUT8-CDG)
- G6PC3-congenital disorder of glycosylation (G6PC3-CDG), Severe congenital neutropenia (SCN)
- Hyperphosphatemic familial tumoral calcinosis (HFTC), Congenital disorders of glycosylation (GALNT3-CDG)
- Polycystic kidney disease (PKD), Congenital disorders of glycosylation (GANAB-CDG)
- GFPT1-congenital disorder of glycosylation (GFPT1-CDG), Congenital myasthenic syndrome 12 (CMS12)
- Childhood-onset spasticity with hyperglycinemia (SPAHGC)
- Glutamine synthetase deficiency
- GM2-gangliosidosis, AB variant
- GMPPA-congenital disorder of glycosylation (GMPPA-CDG), Alacrima, achalasia and intellectual disability syndrome (AAID)
- GMPPB-related conditions:
    - Muscular dystrophy-dystroglycanopathy type A14 (MDDGA14), type B14 (MDDGB14) and type C14 (MDDGC14)
    - Congenital myasthenic syndrome (CMS)
- GNE-related myopathy, Sialuria
- GNPTAB-related conditions:
    - Mucolipidosis type II (ML II) (I-cell disease, Pacman dysplasia)
    - Mucolipidosis type III (ML III) (pseudo-Hurler polydystrophy)
    - Congenital disorders of glycosylation (GNPTAB-CDG)
- Mucopolysaccharidosis type IIID (MPS IIID) (Sanfilippo syndrome D)
- Geroderma osteodysplastica, Congenital disorders of glycosylation (GORAB-CDG)
- Myoclonic epilepsy; GOSR2-congenital disorder of glycosylation (GOSR2-CDG)
- GPAA1-congenital disorder of glycosylation (GPAA1-CDG)
- Tay-Sachs disease
- Sandhoff disease
- HGSNAT-related conditions:
    - Mucopolysaccharidosis IIIC (MPS IIIC) (Sanfilippo syndrome C)
    - Retinitis pigmentosa (RP)

- 3-hydroxy-3-methylglutaryl (HMG)-CoA synthase deficiency
- Multiple mitochondrial dysfunctions syndrome 3 (MMDS3)
- Multiple mitochondrial dysfunctions syndrome 4 (MMDS4)
- Severe congenital neutropenia (SCN) due to JAGN1 deficiency, Congenital disorders of glycosylation (JAGN1-CDG)
- Neuronal ceroid lipofuscinosis type 14 (CLN14) (Progressive myoclonic epilepsy with or without intracellular inclusions)
- Muscular dystrophy-dystroglycanopathy type A6 (MDDGA6) and B6 (MDDGB6)
- Spondylocostal dysostosis, Congenital disorders of glycosylation (LFNG-CDG)
- Hyperglycinemia, lactic acidosis and seizures (HGCLAS), Pyruvate dehydrogenase lipoic acid synthetase deficiency (PDHLD)
- Lipoyltransferase 1 deficiency
- Neonatal encephalopathy with lactic acidosis and brain anomalies (NELABA)
- Immunodeficiency with magnesium defect, Epstein-Barr virus infection, and neoplasia (XMEN), Congenital disorders of glycosylation (MAGT1-CDG)
- MAN1B1-congenital disorder of glycosylation (MAN1B1-CDG)
- Neuronal ceroid lipofuscinosis type 7 (CLN7), Retinal dystrophy
- MGAT2-congenital disorder of glycosylation (MGAT2-CDG, CDG-IIa)
- MOGS-congenital disorder of glycosylation (MOGS-CDG, CDG-IIb)
- MPDU1-congenital disorder of glycosylation (MPDU1-CDG, CDG-If)
- MPI-Congenital disorder of glycosylation (CDG-Ib)
- Mucopolysaccharidosis type IIIB (MPS IIIB) (Sanfilippo syndrome B)
- NANS-congenital disorder of glycosylation (NANS-CDG), Spondyloepimetaphyseal dysplasia syndrome
- Multiple mitochondrial dysfunctions syndrome 1 (MMDS1)
- NGLY1-congenital disorder of glycosylation (CDG-1V)
- Niemann-Pick disease type C (NPC)
- Early infantile epileptic encephalopathy (EIEE), NUS1-congenital disorder of glycosylation (NUS1-CDG, CDG-Iaa)
- OGT-related conditions:
    - X-linked intellectual disability (XLID)
    - Neurodevelopmental seizure condition
    - Congenital disorders of glycosylation (OGT-CDG)
- Succinyl CoA:3-oxoacid CoA transferase (SCOT) deficiency
- Brachyolmia, Congenital disorders of glycosylation (PAPSS2-CDG)
- PGAP1-related intellectual disability
- PGAP2-congenital disorder of glycosylation (PGAP2-CDG)
- PGAP3-congenital disorder of glycosylation (PGAP3-CDG)
- PGM3-congenital disorder of glycosylation (PGM3-CDG)
- PIGA-congenital disorder of glycosylation (PIGA-CDG)
- PIGB-congenital disorder of glycosylation (PIGB-CDG) (Early infantile epileptic encephalopathy)
- PIGC-congenital disorder of glycosylation (PIGC-CDG) (Glycosylphosphatidylinositol biosynthesis defect)
- PIGG-congenital disorder of glycosylation (PIGG-CDG)
- PIGL-congenital disorder of glycosylation (PIGL-CDG) (CHIME syndrome)
- PIGN-congenital disorder of glycosylation (PIGN-CDG) (Multiple congenital anomalies-hypotonia-seizures syndrome)

- PIGO-congenital disorder of glycosylation (PIGO-CDG), Hyperphosphatasia with intellectual disability syndrome
- PIGP-congenital disorder of glycosylation (PIGP-CDG), (Developmental and epileptic encephalopathy, Early infantile epileptic encephalopathy)
- PIGQ-congenital disorder of glycosylation (PIGQ-CDG) (Early infantile epileptic encephalopathy)
- PIGT-congenital disorder of glycosylation (PIGT-CDG) (Glycosylphosphatidylinositol biosynthesis defect 7)
- PIGU-congenital disorder of glycosylation (PIGU-CDG) (Glycosylphosphatidylinositol biosynthesis defect 21)
- PIGV-congenital disorder of glycosylation (PIGV-CDG), Hyperphosphatasia with intellectual disability syndrome, Mabry syndrome
- PIGW-congenital disorder of glycosylation (PIGW-CDG), Glycosylphosphatidylinositol (GPI) biosynthesis defect 11, Hyperphosphatasia with intellectual disability syndrome 5
- Glycosylphosphatidylinositol biosynthesis defect (PIGM-CDG)
- PMM2-congenital disorder of glycosylation (CDG-Ia)
- Dowling-Degos disease 2 (DDD2), POFUT1-congenital disorder of glycosylation (POFUT1-CDG)
- Dowling-Degos disease (DDD), Limb-girdle muscular dystrophy type R21 (LGMDR21)
- Muscular dystrophy-dystroglycanopathy type A3, B3,C3
- Muscular dystrophy-dystroglycanopathy type A8 and C8
- Muscular dystrophy-dystroglycanopathy type A12, C12
- Muscular dystrophy-dystroglycanopathy type A1, B1, C1
- Muscular dystrophy-dystroglycanopathy type A2, B2, C2
- Neuronal ceroid lipofuscinosis 1 (CLN1)
- Polycystic liver disease (PCLD), Congenital disorders of glycosylation (PRKCSH-CDG)
- RFT1-congenital disorder of glycosylation (RFT1-CDG, CDG-In)
- RPN2-congenital disorder of glycosylation (RPN2-CDG)
- Muscular dystrophy-dystroglycanopathy type A10 (MDDGA10)
- Chylomicron retention disease (CMRD), Congenital disorders of glycosylation (SAR1B-CDG)
- Craniolenticulosutural dysplasia (CLSD), Congenital disorders of glycosylation (SEC23A-CDG)
- SEC23B-congenital disorder of glycosylation (SEC23B-CDG), Congenital dyserythropoietic anemia, type II (CDAII)
- Cole-Carpenter syndrome, Congenital disorders of glycosylation (SEC24D-CDG)
- Polycystic liver disease 2 (PCLD2), Congenital disorders of glycosylation (SEC63-CDG)
- Mucopolysaccharidosis type IIIA (MPS IIIA) (Sanfilippo syndrome A)
- SLC10A7-congenital disorder of glycosylation (SLC10A7-CDG) (Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis syndrome)
- SLC16A1-related conditions
- SLC26A2-related conditions:
    - Achondrogenesis, type IB (ACG1B)
    - Atelosteogenesis type 2 (AO2)
    - Diastrophic dysplasia (DTD)
    - Multiple epiphyseal dysplasia 4 (EDM4)
- SLC35A1-congenital disorder of glycosylation (SLC35A1-CDG, CDG-IIf)
- Congenital disorder of glycosylation (SLC35A2-CDG, CDG-IIm)

- SLC35A3-congenital disorder of glycosylation (SLC35A3-CDG)
- SLC35C1-congenital disorder of glycosylation (SLC35C1-CDG, CDG-IIc), Leukocyte adhesion deficiency type II
- Schneckenbecken dysplasia (SBD), Congenital disorders of glycosylation (SLC35D1-CDG)
- Glycogen storage disease Ib (GSD Ib)
- SLC39A8-congenital disorder of glycosylation (SLC39A8-CDG, CDG-IIn)
- Glycine encephalopathy with normal serum glycine
- Lysinuric protein intolerance (LPI)
- SLC9A7-related X-linked intellectual disability, Congenital disorder of glycosylation (SLC9A7-CDG)
- SRD5A3-congenital disorder of glycosylation (CDG-Iq) (Kahrizi syndrome)
- Congenital disorders of glycosylation (SSR3-CDG)
- SSR4-Congenital Disorder of Glycosylation (CDG type 1y)
- Congenital disorders of glycosylation (ST3GAL3-CDG) (Developmental and epileptic encephalopathy, Early infantile epileptic encephalopathy)
- ST3GAL5-congenital disorder of glycosylation (ST3GAL5), GM3 synthase deficiency
- STT3A-congenital disorder of glycosylation (CDG-Iw)
- STT3B-congenital disorder of glycosylation (STT3-CDG, CDG-Ix)
- Catel-Manzke syndrome, Congenital disorders of glycosylation (TGDS-CDG)
- Congenital disorder of glycosylation (CDG-IIk)
- TMEM199-congenital disorder of glycosylation (TMEM199-CDG, CDG-IIp)
- Neuronal ceroid lipofuscinosis 2 (CLN2)
- Early infantile epileptic encephalopathy (EIEE), Congenital disorders of glycosylation (TRAK1-CDG)
- TRAPC11-congenital disorder of glycosylation (TRAPC11-CDG), Limb-girdle muscular dystrophy type 2S
- Progressive encephalopathy with brain atrophy and spasticity (PEBAS), Congenital disorders of glycosylation (TRAPPC12-CDG)
- Spondyloepiphyseal dysplasia tarda (SEDT), Congenital disorders of glycosylation (TRAPPC2-CDG)
- Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy (NEDMEBA); Congenital disorders of glycosylation (TRAPPC6B-CDG)
- TRAPPC9-related intellectual disability, Congenital disorders of glycosylation (TRAPPC9-CDG)
- Achondrogenesis Type 1A, Odontochondrodysplasia (ODCD), Congenital disorders of glycosylation (TRIP11-CDG)
- TUSC3-congenital disorder of glycosylation (TUSC3-CDG)
- X-linked myopathy with excessive autophagy, Congenital disorders of glycosylation (VMA21-CDG)
- Cohen syndrome, Congenital disorders of glycosylation (VPS13B-CDG)
- Desbuquois dysplasia type 2, Baratela-Scott syndrome (BSS), Congenital disorders of glycosylation (XYLT1-CDG)
- Spondyloocular syndrome, Congenital disorders of glycosylation (XYLT2-CDG)