Genomic Corporation

Decoding the Human Genome

Decoding the Human Genome in partnership with world leading Biotech

TEST CATALOG

in association with Centogene, Germany

CENTOGENE

THE RARE DISEASE COMPANY

About Genomics

Genomic Corporation offers comprehensive, high quality genetic testing services in Pakistan in association with the world's leading Biotech, **Centogene Germany**. We provide a whole range of advanced testing starting from a single gene to the whole genome sequencing and are resolute in our purpose of revolutionizing genetic diagnostics in Pakistan by providing fast, accurate and reliable results.

About Centogene

CENTOGENE is an internationally accredited and worldwide leader in the field of genetic diagnostics for rare hereditary diseases - with the largest test portfolio worldwide. Testing samples from over 110 different countries allows CENTOGENE a unique insight into the epidemiological basis of hereditary disorders, which is crucial for interpreting results and transform it into medical reports. CENTOGENE has developed a comprehensive mutation database that is pivotal to offering high-quality diagnostic reporting and medical interpretation; thoroughly interpreting each patient's sequence data.

Why Choose Centogene

Centogene provides:

- The best interpretation of genetic test results and provides expert reviews of patient genetic condition for all types of genetic counseling
- Advanced logistical solutions from sample sending to report administration
- Global genotype-phenotype database for rare diseases
- Shortest turnaround time and fast reporting
- Offers more than 2.900 single gene tests and >200 NGS panels designed according to focused medical needs, which are capable of detecting all potential disease-causing genetic changes ("mutations")
- Technical specificity for methods in use is > 99,9% and technical sensitivity > 99,8%

CLIA #99D2049715

PANELS

SR NO	TEST PANELS	GENES
1	Abnormal mineralization panel	ALPL, ANKH, AP2S1, CASR, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FGF23, GNA11, PHEX, PTH1R, SLC34A1, SLC34A3, SLC9A3R1, VDR
2	Achromatopsia panel	ATF6, CNGA3, CNGB3, CNNM4, GNAT2, OPN1LW, OPN1MW, PDE6C, PDE6H, RPGR
3	Afibrinogenemia panel	FGA, FGB, FGG
4	Agammaglobulinemia panel	BLNK, BTK, CD79A, CD79B, IGHM, IGLL1, LRRC8A, PIK3R1, SH2D1A
5	Aicardi-Goutieres syndrome panel	ADAR, IFIH1, TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1
6	Albinism panel	AP3B1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, EDN3, EDNRB, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1
7	AllNeuro panel	A2M, AAAS, AANAT, AARS, AARS2, AASS, ABAT, ABCA1, ABCB7, ABCC6, ABCC8, ABCD1, ABCD4, ABHD12, ABHD5, ACACA, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACKR1, ACO2, ACOX1, ACSF3, ACSL4, ACTA1, ACTA2, ACTB, ACTG1, ACVRL1, ACY1, ADAM10, ADAR, ADCY5, ADGRG1, ADGRV1, ADK, ADNP, ADSL, AFF2, AFG3L2, AGA, AGK, AGL, AGRN, AGXT, AHCY, AHI1, AIFM1, AIMP1, AKT1, AKT3, ALAD, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX5AP, ALS2, ALX1, ALX3, ALX4, AMACR, AMN, AMPD1, AMPD2, AMT, ANG, ANK3, ANKRD11, ANO10, ANO3, ANO5, AP1S1, AP1S2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APOE, APP, APTX, AR, ARFGEF2, ARG1, ARHGAP31, ARHGEF10, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARL13B, ARL6, ARSA, ARSB, ARSE, ARSI, ARX, ASAH1, ASCL1, ASL, ASNS, ASPA, ASPM, ASS1, ASXL1, ASXL3, ATCAY, ATIC, ATL1, ATM, ATP13A2, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B3, ATP2B4, ATP5E, ATP6AP2, ATP6V0A2, ATP7A, ATP7B, ATP8A2, ATPAF2, ATR, ATRX, ATXN3, B3GLCT, B4GALNT1, B4GALT1, B9D1, B9D2, BAG3, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCOR, BCS1L, BDNF, BEST1, BICD2, BIN1, BLOC1S3, BLOC1S6, BOLA3, BRAF, BRAT1, BRWD3, BSCL2, BSND, C10orf2, C12orf57, C12orf65, C19orf12, C5orf42, C9orf72, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1H, CACNA1S, CACNB4, CACNG2, CAMTA1, CAPN3, CASC5, CASK, CASR,

		CAV3, CBL, CC2D1A, CC2D2A, CCDC28B, CCDC78, CCDC88C, CCM2, CCT5, CD207, CD320, CD36, CD59, CD96, CDH15, CDK11A, CDK5RAP2, CDKL5, CDON, CENPJ, CEP135, CEP152, CEP290, CEP41, CEP63, CERS1, CFL2, CHAT, CHCHD10, CHD2, CHD7, CHD8, CHMP1A, CHMP2B, CHRM3, CHRNA1, CHRNA2, CHRNA4, CHRNB1, CHRNB2, CHRND, CHRNE, CHRNG, CHSY1, CISD2, CISH, CIZ1, CLCN1, CLCN2, CLCNKA, CLCNKB, CLIC2, CLN3, CLN5, CLN6, CLN8, CNBP, CNTN1, CNTNAP2, CNTNAP4, COA5, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL12A1, COL18A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COQ2, COQ8A, COQ9, COX10, COX15, COX6A1, COX6B1, CP, CPA6, CPT2, CR1, CRADD, CRBN, CREBBP, CRYAB, CSF1R, CSF2RB, CSPP1, CST3, CSTB, CTC1, CTDP1, CTNNB1, CTSD, CUL4B, CWF19L1, CYP11B2, CYP27A1, CYP2U1, CYP7B1, DAG1, DARS2, DBT, DCAF17, DCTN1, DCX, DDHD1, DDHD2, DDOST, DEPDC5, DES, DGUOK, DHCR7, DHH, DHTKD1, DIAPH3, DKC1, DLD, DLG3, DMD, DNAH9, DNAJB2, DNAJB6, DNAJC19, DNAJC5, DNAJC6, DNM1L, DNM2, DNMT1, DOCK8, DOK7, DOLK, DPAGT1, DPM1, DPM3, DRD2, DRD3, DSC3, DST, DTNBP1, DYNC1H1, DYNC2H1, DYRK1A, DYSF, EARS2, EBP, EDN3, EDNRB, EEF2, EFHC1, EFTUD2, EGR2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4E, EIF4G1, ELK1, ELOVL4, ELOVL5, EMD, EMX2, ENO3, ENTPD1, EOMES, EP300, EPB41L1, EPM2A, ERBB4, ERCC1, ERCC2, ERCC5, ERCC6, ERLIN1, ERLIN2, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EXOC8, EXOSC3, EXOSC8, F2, F5, FA2H, FADD, FAM126A, FAM134B, FANCB, FASTKD2, FBLN5, FBXO38, FBXO7, FCGR2B, FGA, FGD1, FGD4, FGF10, FGF14, FGFR1, FGFR2, FGFR3, FH, FHL1, FIG4, FKRP, FKTN, FLNA, FLNC, FLRT1, FLVCR1, FLVCR2, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FOXP3, FOXRED1, FREM1, FRG1, FRMD7, FTL, FTO, FTSJ1, FUS, FXN, FZD9, GAA, GABRA1, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNS, GAMT, GAN, GARS, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCSH, GDAP1, GDI1, GDNF, GFAP, GFER, GFM1, GFPT1, GIGYF2, GJB1, GJB3, GJC2, GK, GLB1, GLDC, GLI2, GLI3, GLRA1, GLRB, GM2A, GMPPB, GNAL, GNB4, GNE, GNPAT, GNS, GOSR2, GP1BA, GPC3, GPR143, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIP1, GRM1, GRN, GSN, GUSB, GYG1, GYS1, HADHA, HADHB, HCCS, HDAC4, HDAC8, HEPACAM, HERC2, HESX1, HEXA, HEXB, HFE, HGSNAT, HINT1, HK1, HMGCS2, HNRNPDL, HOXA1, HOXD10, HPCA, HPD, HPRT1, HPS1, HPS3, HPS4, HPS5, HPS6, HPSE2, HRAS, HSD17B10, HSD17B4, HSPB1, HSPB3, HSPB8, HSPD1, HTRA1, HTRA2, HTT, HUWE1, HYAL1, ICAM1, ICK, IDS, IDUA, IER3IP1, IFRD1, IFT140, IFT27, IGBP1, IGF1, IGHMBP2, IKBKAP, IKBKG, IL11RA, IL1RAPL1, IL1RN, INF2, INPP5E, INS, IQSEC2, IRX5, ISPD, ITM2B, ITPR1, JRK, KANK1, KARS, KAT6B, KBTBD13, KCNA1, KCNC3, KCND3, KCNE1L, KCNJ1, KCNJ10, KCNJ11, KCNK18, KCNK9, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, KDM6A, KIAA0196, KIAA2022, KIF11, KIF1A, KIF1B, KIF1C,

		KIF21A, KIF5A, KIF7, KIRREL3, KLF8, KMT2A, KMT2D, KRAS, KRIT1, KRT5, L1CAM, LAMA2, LAMB1, LAMB2, LAMC3, LAMP2, LARGE, LBR, LDB3, LDHA, LGI1, LHX4, LIMS2, LINS, LITAF, LMBRD1, LMNA, LMNB1, LPIN1, LPIN2, LRP2, LRPPRC, LRRK2, LRSAM1, LYZ, LZTFL1, MAG, MAGEL2, MAGI2, MAGT1, MAMLD1, MAN1B1, MAOA, MAPK10, MAPT, MARS, MARS2, MASP1, MATR3, MBD5, MBTPS2, MCEE, MCPH1, MECP2, MED12, MED17, MED23, MED25, MEF2C, MET, MFN2, MFSD8, MGAT2, MID1, MKKS, MKS1, MLC1, MMAA, MMAB, MMACHC, MMADHC, MOGS, MPDU1, MPDZ, MPI, MPV17, MPZ, MRE11A, MSX1, MSX2, MTFMT, MTHFR, MTM1, MTMR14, MTMR2, MTO1, MTOR, MTPAP, MTR, MTRR, MUSK, MUT, MVK, MYCN, MYF6, MYH14, MYH7, MYH9, MYO5A, MYOT, NAA10, NAGA, NAGLU, NAT8L, NBN, NDE1, NDP, NDRG1, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NEDD4L, NEFH, NEFL, NEU1, NF1, NFIX, NFU1, NGF, NHEJ1, NHLRC1, NHS, NIPA1, NIPBL, NKX2-1, NLGN3, NLGN4X, NLRP12, NLRP3, NOD2, NOG, NOL3, NOP56, NOS2, NOS3, NOTCH2, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NRAS, NRG1, NRXN1, NSD1, NSDHL, NSUN2, NT5C2, NTRK1, NTRK2, NUBPL, NXF5, OCLN, OCRL, OFD1, OPA1, OPA3, OPHN1, OPTN, ORC1, OTC, PAFAH1B1, PAH, PAK3, PANK2, PARK2, PARK7, PAX6, PC, PCBD1, PCDH19, PCK2, PCNT, PDCD10, PDE6D, PDE8B, PDHA1, PDK3, PDSS1, PDSS2, PDYN, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PFN1, PGAM2, PGAP1, PGK1, PGM1, PHF6, PHF8, PHKA1, PHOX2B, PHYH, PIEZO2, PIGA, PIGL, PIGN, PIGO, PIGV, PIK3CA, PIK3R2, PIK3R5, PINK1, PLA2G6, PLCB1, PLEC, PLEKHG4, PLEKHG5, PLP1, PMM2, PMP22, PNKD, PNKP, PNPLA2, PNPLA6, PNPO, POGZ, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PORCN, PPT1, PQBP1, PREPL, PRICKLE1, PRICKLE2, PRKAG2, PRKCG, PRKCH, PRKRA, PRNP, PROP1, PRPH2, PRPS1, PRRT2, PRRX1, PRSS12, PRX, PSAP, PSEN1, PSEN2, PTCH1, PTEN, PTF1A, PTPN11, PTS, PUS1, PVRL1, PYGM, QARS, QDPR, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAB40AL, RAB7A, RABGGTA, RAD21, RAD50, RAF1, RAI1, RANBP2, RAPSN, RARS2, RBBP8, RBFOX1, RBFOX3, RBM10, RBM8A, REEP1, REEP2, RELN, RET, RFT1, RIN2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF135, RNF170, RNU4ATAC, ROGDI, ROR2, RPGRIP1L, RPL10, RPS6KA3, RRM2B, RTN2, RUBCN, RYR1, SACS, SAMHD1, SBF1, SBF2, SCARB2, SCN10A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN5A, SCN8A, SCN9A, SCO1, SCO2, SCP2, SDCCAG8, SDHA, SDHAF1, SEPN1, SEPSECS, SERPINI1, SETBP1, SETD2, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SGSH, SH3TC2, SHANK2, SHANK3, SHH, SHOC2, SHROOM4, SIGMAR1, SIL1, SIX3, SIX6,

		SKI, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A3, SLC20A2, SLC22A5, SLC25A12, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A4, SLC2A1, SLC30A10, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC3A1, SLC4A1, SLC4A4, SLC5A7, SLC6A1, SLC6A3, SLC6A5, SLC6A8, SLC9A6, SLC9A9, SMARCA2, SMARCA4, SMARCB1, SMC1A, SMC3, SMCHD1, SMN1, SMN2, SMPD1, SMS, SNAP29, SNCA, SNCAIP, SNCB, SNIP1, SNX3, SOBP, SOD1, SORL1, SOS1, SOX10, SOX3, SPAST, SPG11, SPG20, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SPTLC1, SPTLC2, SRD5A3, SRPX2, ST3GAL3, ST3GAL5, STIL, STRADA, STUB1, STXBP1, SUCLA2, SUCLG1, SUMF1, SURF1, SYN1, SYNE1, SYNE2, SYNGAP1, SYNJ1, SYP, SYT14, SZT2, TACO1, TAF1, TAF2, TARDBP, TAS2R38, TAZ, TBC1D24, TBCE, TBL1XR1, TBX1, TCAP, TCF4, TCTN1, TCTN2, TCTN3, TDP1, TECPR2, TECR, TFAP2A, TFAP2B, TFG, TG, TGFB1, TGFB2, TGFBR1, TGFBR2, TGIF1, TGM6, TH, THAP1, TICAM1, TIMM8A, TIRAP, TK2, TLR3, TLR5, TMCO1, TMEM126A, TMEM138, TMEM165, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TMLHE, TNF, TNFSF4, TNNT1, TNPO3, TOR1A, TP63, TPK1, TPM2, TPM3, TPP1, TRAF3, TRAPPC11, TRAPPC9, TREM2, TREX1, TRIM2, TRIM32, TRPM6, TRPM7, TRPS1, TRPV4, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TTBK2, TTC19, TTC21B, TTC8, TTI2, TTN, TTPA, TTR, TUBA1A, TUBA4A, TUBA8, TUBB2B, TUBB3, TUBB4A, TUBGCP6, TUSC3, TWIST1, TYMP, TYROBP, UBA1, UBE2A, UBE3A, UBQLN2, UCHL1, UMPS, UNC93B1, UPB1, UPF3B, UQCRB, UQCRQ, USP8, USP9X, VAMP1, VANGL1, VAPB, VAX1, VCP, VDAC1, VEGFA, VHL, VIPAS39, VLDLR, VPS13A, VPS13B, VPS35, VPS37A, VRK1, WAC, WDPCP, WDR45, WDR48, WDR62, WDR81, WFS1, WNK1, WNT10A, WNT3, WNT5A, WNT7A, WWOX, XBP1, XK, YAP1, YARS, YWHAE, ZBTB16, ZBTB18, ZCCHC12, ZDHHC15, ZDHHC9, ZEB2, ZFR, ZFYVE26, ZFYVE27, ZIC2, ZIC3, ZNF335, ZNF423, ZNF592, ZNF711, ZNF81
8	Alport syndrome panel	COL4A3, COL4A4, COL4A5
9	Alzheimer dementia and dementia panel	APOE, APP, PRNP, PSEN1, PSEN2, SORL1, TREM2
10	Amyotrophic lateral sclerosis (ALS) panel	ALS2, ANG, CHCHD10, CHMP2B, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, MATR3, NEFH, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TUBA4A, UBQLN2, VAPB, VCP
11	Arrhythmia, hereditary panel	AKAP9, ANK2, CACNA1C, CACNB2, CASQ2, CAV3, DSC2, DSG2, DSP, GPD1L, JUP, KCNA5, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNQ1, NPPA, PKP2, PLN, RYR2, SCN1B, SCN3B, SCN4B, SCN5A, SNTA1, TGFB3, TMEM43

12	Arrhythmogenic right ventricular cardiomyopathy panel	DSP, DSG2, DSC2, JUP, PKP2, RYR2, TGFB3, TMEM43
13	Arthrogryposis panel	TPM2, MYBPC1, MYH3, TNNT3, TNNI2, MYH8, FBN2, PIEZO2, ECEL1, DOK7, RAPSN
14	Ashkenazi panel (advanced)	ABCC8 (c.3989-9G>A, p.F1387del (NM_000352.3)), ASPA (c.433-2A>G, p.Y231X, p.E285A, p.A305E (NM_001128085.1)), BCKDHB (c.548G>C, c.832G>A, c.1114G>T (NM_183050.2)), BLM (c.2207_2212delATCTGAinsTAGATTC (NM_000057.2)), BRCA1 (c.68_69delAG, c.5266dupC (NM_007294.3)), BRCA2 (c.5946delT (NM_000059.3 2)), CFTR (c.1521_1523delCTT, c.1519_1521delATC, c.3454G>C, c.3846G>A, c.3718-2477C>T, c.3909C>G, c.3808G>A, c.1040G>A, c.1865G>A, c.489+1G>T, c.1657C>T, c.262_263del, c.1652G>A, c.2988+1G>A, c.350G>A, c.579+1G>T, c.1000C>T, c.1679G>C, c.3484C>T, c.3528delC, c.254G>A, c.1364C>A, c.2052del, c.1766+1G>A, c.443T>C, 1078delT, c.1624G>T, c.2657+5G>A, c.1585-1G>A, c.178G>T (NM_000492.3)), CLRN1 (c.144T>G (NM_174878.2)), DLD (c.104dupA, c.685G>T (NM_000108.3)), FANCC (c.67delG, c.456+4A>T, NM_000136.2), G6PC (c.247C>T (NM_000151.3)), GBA (c.84dupG, c.115+1G>A, c.1448T>C, c.1226A>G, c.1604G>A, c.1297G>T (NM_001005741.2)), HEXA (c.1274_1277dupTATC, c.1421+1G>C, c.805G>A, c.739C>T, c.745C>T (NM_000520.4)), IKBKAP (c.2204+6T>C, c.2087G>C (NM_003640.3)), MCOLN1 (c.406-2A>G, 6.4kb del/ g.511_6943del (exon 01 to part exon 07) (NM_020533.2)), NEB (p.R2478_D2512del/ c.7431+1917_7536+372del (exon 55 deletion))(NM_001271208.1)), PCDH15 (c.733C>T, (NM_033056.3)), SMPD1 (c.996delC, c.911T>C, c.1493G>, c.1829_1831delGCC (NM_000543.4)), PKHD1 (c.3761 (NM_138694.3)), TMEM216 (c.218G>T, (NM_001173991.2)), MPL (c.79+2T>A (NM_005373.2), CPT2 (c.338C>T, c.1239_1240del, c.1342T>C (NM_000098.2)), PMM2 (c.422G>A, c.338C>T, c.357C>A, c.691G>A (NM_000303.2)), FAH (c.782C>T (NM_000137.2)), FKTN (c.1167dup (NM_001079802.1)), ATP7B (c.3191A>C, c.3207C>A, c.2333G>T, c.1934T>G (NM_000053.2))
15	Ashkenazi panel (basic)	ABCC8 (c.3989-9G>A, p.F1387del (NM_000352.3)), ASPA (p.Y231X, p.E285A, p.A305E (NM_001128085.1)), BCKDHB (c.548G>C, c.832G>A, c.1114G>T (NM_183050.2)), BLM (c.2207_2212delATCTGAinsTAGATTC (NM_000057.2)), BRCA1 (c.68_69delAG, c.5266dupC (NM_007294.3)), BRCA2 (c.5946delT (NM_000059.3 2)), CFTR (c.1521_1523delCTT, c.1519_1521delATC, c.3454G>C, c.3846G>A, c.3718-2477C>T, c.3909C>G, c.2988+1G>A, c.350G>A, c.1000C>T, c.254G>A, c.1364C>A, c.2052del, c.1624G>T, c.1585-1G>A (NM_000492.3)), DLD

		(c.104dupA, c.685G>T (NM_000108.3)), FANCC (c.456+4A>T, NM_000136.2), G6PC (c.247C>T (NM_000151.3)), GBA (c.84dupG, c.115+1G>A, c.1448T>C, c.1226A>G, c.1604G>A, c.1297G>T (NM_001005741.2)), HEXA (c.1274_1277dupTATC, c.1421+1G>C, c.805G>A, c.739C>T, c.745C>T (NM_000520.4)), IKBKAP (c.2204+6T>C (NM_003640.3)), MCOLN1 (c.406-2A>G, 6.4kb del/ g.511_6943del (exon 01 to part exon 07) (NM_020533.2)), SMPD1 (c.996delC, c.911T>C, c.1493G>T, c.1829_1831delGCC (NM_000543.4)), TMEM216 (c.218G>T, (NM_001173991.2))
16	Atypical hemolytic uremic syndrome panel	ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, DGKE, MMACHC, PIGA, PLG, THBD
17	Autoimmune lymphoproliferative syndrome panel	CASP10, CASP8, CTLA4, FADD, FAS, FASLG, ITK, KRAS, MAGT1, NRAS, PRKCD

18	B-negative SCID panel	ADA, AK2, DCLRE1C, LIG4, NHEJ1, RAC2, RAG1, RAG2
19	B-positive SCID panel	CD3D, CD3E, CD247, FOXN1, IL2RG, IL7R, JAK3, ORAI1, PNP, PTPRC, RMRP, STAT5B, STIM1, TBX1, ZAP70
20	Bardet Biedl panel	ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, IFT27, LZTFL1, MKKS, MKS1, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP
21	Bartter Syndrome panel	ATP6V1B1, BSND, CA2, CASR, CLCNKA, CLCNKB, CLDN16, CLDN19, FXYD2, HSD11B2, KCNJ1, KCNJ10, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A2, SLC12A3, SLC4A1, SLC4A4, WNK1, WNK4
22	Bethlem myopathy panel	COL6A1, COL6A2, COL6A3, COL12A1
23	Bone marrow failure panel	ABCB7, AK2, ALAS2, ANKRD26, BRCA2, BRIP1, CASP10, CBL, CDAN1, CSF3R, CXCR4, DKC1, ELANE, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GFI1, HAX1, HOXA11, JAGN1, KLF1, LYST, MASTL, MPL, MYH9, NBN, NHP2, NOP10, PALB2, PARN, PRF1, PUS1, RAC2, RAD51C, RPL11, RPL15, RPL26, RPL27, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RTEL1, SBDS, SEC23B, SLC19A2, SLC25A38, SLC46A1, SLX4, SRP72, STX11, STXBP2, TCN2, TERC, TERT, TINF2, UBE2T, VPS45, WAS, WRAP53, XRCC2

24	Brain iron accumulation syndromes panel	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, SCP2, WDR45
25	BRCA1, BRCA2 panel	BRCA1, BRCA2
26	BRCA1, BRCA2 somatic mutation analysis	BRCA1, BRCA2
27	Breast ovarian cancer panel	CDH1, PTEN, STK11, TP53
28	Breast ovarian cancer panel PLUS	ATM, BARD1, BRIP1, CHEK2, MEN1, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS1, PMS2, RAD50, RAD51C, RAD51D, XRCC2
29	Brugada syndrome panel	CACNA1C, CACNB2, GPD1L, HCN4, KCNE3, SCN1B, SCN3B, SCN5A, SLMAP

30	Cancer Hotspot Panel	covering most frequent mutations in genes ABL1, AKT1, ALK, APC, ATM, BRAF, CDH1, CDKN2A, CSF1R, CTNNB1, EGFR, ERBB2, ERBB4, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, GNA11, GNAQ, GNAS, HNF1A, HRAS, IDH1, IDH2, JAK2, JAK3, KDR, KIT, KRAS, MET, MLH1, MPL, NOTCH1, NPM1, NRAS, PDGFRA, PIK3CA, PTEN, PTPN11, RB1, RET, SMAD4, SMARCB1, SMO, SRC, STK11, TP53, VHL
31	Cardiomyopathy dilated panel	ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FKTN, GATA4, GATAD1, ILK, LAMA4, LAMP2, LDB3, LMNA, MURC, MYBPC3, MYH6, MYH7, MYPN, NEBL, NEXN, PDLIM3, PKP2, PLN, PRDM16, RAF1, RBM20, SCN5A, SGCD, TAZ, TBX20, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL
32	Cardiomyopathy hypertrophic panel	ACTC1, ACTN2, ANKRD1, CALR3, CAV3, CRYAB, CSRP3, DES, FHL2, FLNC, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYPN, NEXN, PDLIM3, PLN, PRKAG2, SLC25A4, SOS1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL
33	Cataract panel	AGK, BCOR, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, EPHA2, EYA1, FOXC1, FOXE3, FTL, FYCO1, GALK1, GCNT2, GJA3, GJA8, HSF4, LEMD2, LIM2, LSS, MAF, MIP, NHS, P3H2, PAX6, PITX3, SIPA1L3, SLC16A12, TDRD7, UNC45B, VIM, VSX2, WFS1

34	Catecholaminergic polymorphic ventricular tachycardia panel	RYR2, CASQ2, KCNJ2
35	CentoArray Cyto™ 750K	Genome-wide aCGH with 750,000 markers
36	CentoArray Cyto™ HD	Genome-wide aCGH with 2.5 million markers
37	CentoBreast® panel	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, NBN, PALB2, PTEN, RAD51C, STK11, TP53
38	CentoCancer panel	APC, ATM, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, FH, FLCN, MLH1, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SMAD4, STK11, TP53
39	CentoColon extended panel	APC, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53
40	CentoDx Plus™ Solo	Targets exonic regions of ~6700 genes associated with known clinical phenotypes
41	CentoDx Plus™ Trio	Targets exonic regions of ~6700 genes associated with known clinical phenotypes
42	CentoDx Plus™ Trio Plus	Targets exonic regions of ~6700 genes associated with known clinical phenotypes
43	CentoICU™ platinum	AARS, AARS2, AASS, ABAT, ABCA12, ABCA3, ABCB11, ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACTA1, ADA, ADAMTS13, ADAR, ADK, ADNP, ADSL, AGK, AGL, AGPAT2, AGRN, AGXT, AHCY, AICDA, AIFM1, AIMP1, AKAP9, AKR1D1, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG14, ALG2, ALG3, ALG6, ALMS1, ALOX12B, ALOXE3, ALPL, ALS2, AMACR, AMT, ANK1, ANKRD26, ANKS6, ANTXR1, AP2S1, AP4B1, AP4E1, AP4M1, AP4S1, APOB, ARG1, ARL6, ARSA, ARSB, ARX, ASL, ASNS, ASPA, ASPM, ASS1, ATP1A3, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATR, ATRX, AUH, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BDNF, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSND, BTD, BTK, C10orf2, C12orf65, C21orf59, CA12, CACNA1C, CACNB2, CALM1, CAMTA1, CASK, CASR, CAST, CAV3, CBS, CCDC103, CCDC114, CCDC78, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDK5RAP2, CDKL5, CDKN1C, CENPJ, CEP152, CEP290, CERS3, CFH, CFHR3, CFL2, CFTR, CHAT, CHD7, CHKB, CHM, CHRNA1, CHRNB1, CHRND, CHRNE, CIDEC, CLCNKA, CLCNKB, CLPB, CNTN1, COA5, COL11A1, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COMP, COQ2, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CPS1, CPT1A, CPT2, CR2, CRTAP, CTNS, CTPS1, CTSA, CTSD, CUL4B,

		CXCR4, CYP11B1, CYP11B2, CYP17A1, CYP4F22, CYP7B1, D2HGDH, DBT, DCLRE1C, DDC, DDOST, DDR2, DECR1, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DIAPH1, DLAT, DLD, DMD, DNA2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC19, DNM2, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM2, DPYD, DRC1, DSP, DST, DUOX2, DUOXA2, DYSF, EDN3, EEF1A2, EGR2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELANE, ENPP1, EPB42, EPCAM, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYA1, EYA4, F10, F11, F13A1, F2, F5, F7, F8, F9, FADD, FAH, FANCA, FANCB, FANCC, FANCD2, FANCL, FARS2, FASTKD2, FBN1, FBP1, FBXL4, FGA, FGB, FGFR2, FGFR3, FGG, FH, FIG4, FKBP14, FKRP, FKTN, FOXC1, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PC2, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GARS, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GFAP, GFM1, GFPT1, GJA1, GJB2, GJB4, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLUD1, GLYCTK, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GP1BA, GP1BB, GP9, GPC3, GPHN, GPSM2, GSS, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HESX1, HEXA, HEXB, HGD, HGF, HIBCH, HLCS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPGD, HRAS, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, ICOS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IGHMBP2, IGLL1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, ISPD, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, IVD, JAG1, JAGN1, JAK3, JAM3, KAT6A, KAT6B, KBTBD13, KCNE1, KCNH1, KCNH2, KCNJ10, KCNJ11, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1B, KLF1, KLHL40, KLHL41, KRAS, KRT5, LAMA2, LAMA3, LAMB3, LAMC2, LAMP2, LAMTOR2, LARS2, LAS1L, LCT, LHX3, LHX4, LIAS, LIG4, LIPA, LIPN, LIPT1, LMBRD1, LMNA, LPIN1, LRBA, LRPPRC, LRRC8A, MAGEL2, MAGT1, MALT1, MAN2B1, MANBA, MAP2K1, MAP2K2, MASTL, MAT1A, MCCC1, MCCC2, MCEE, MCM4, MCPH1, MECP2, MED12, MEF2C, MEGF10, MFN2, MFSD8, MITF, MKKS, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MPC1, MPI, MPL, MPV17, MPZ, MRPL3, MRPL44, MSMO1, MTHFR, MTM1, MTMR14, MTO1, MTR, MTRR, MUSK, MUT, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NDUFV2, NEB, NEFL, NEU1, NEUROG3, NEXN, NFKB2, NFU1, NGF, NHEJ1, NIPAL4, NIPBL, NKX2-1, NKX2-5, NLRC4, NLRP3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NR0B1, NR3C2, NRAS, NSD1, NSDHL, NUBPL, OAT, OCLN, OCRL, OPA3, OPHN1, OPLAH, ORC1, ORC4, OTC, OXCT1, PAFAH1B1, PAH, PAX2, PAX3, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH19, PCNT, PDCD10, PDE10A, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX13, PEX19, PEX7, PGAP1, PHGDH, PHOX2B, PIGA, PIGN,

		PIGT, PIGV, PIK3CD, PKD2, PKHD1, PKLR, PLCB4, PLEC, PLOD1, PLP1, PMM2, PMP22, PNKP, PNP, PNPLA1, PNPO, PNPT1, POGZ, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, PPT1, PRDM16, PRKAG2, PRKDC, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PSAP, PSAT1, PSPH, PTPN11, PTPRC, PTRF, PTRH2, PTS, PURA, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RAC2, RAF1, RAG1, RAG2, RANBP2, RAPSN, RARS2, RB1, RBBP8, RBM8A, RET, RFT1, RFX5, RFX6, RIT1, RMND1, RMRP, RNASEH2C, RNASET2, RNU4ATAC, RORC, RPS19, RRM2B, RYR1, SALL1, SATB2, SBDS, SCN1A, SCN2A, SCN4A, SCN5A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SEPN1, SERAC1, SERPINC1, SERPING1, SFTPB, SFTPC, SFTPD, SHOC2, SIL1, SIX3, SIX5, SKI, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC26A2, SLC26A3, SLC2A1, SLC30A2, SLC33A1, SLC37A4, SLC3A1, SLC46A1, SLC4A1, SLC52A1, SLC52A3, SLC5A1, SLC5A5, SLC6A1, SLC6A3, SLC6A5, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNAI2, SNX10, SOS1, SOX10, SOX2, SOX9, SPAST, SPEG, SPINK5, SPINT2, SPR, SPRED1, SPTA1, SPTAN1, SPTB, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STAT3, STIL, STIM1, STS, STT3B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAT, TAZ, TBC1D24, TBCE, TBX19, TBX5, TCAP, TCN2, TFR2, TG, TGM1, TH, THRA, TJP2, TMCO1, TMEM165, TMEM173, TMEM5, TMEM70, TNFRSF13B, TNFRSF13C, TNFSF4, TNNT1, TP63, TPM2, TPM3, TPO, TPP1, TRIP11, TRMU, TRPV4, TSC1, TSC2, TSFM, TSHB, TSHR, TSPYL1, TTC7A, TTN, TUBA8, TUBB2A, UBA1, UGT1A1, UMPS, UNG, UPB1, UQCRC2, UROD, UROS, WAS, WDPCP, WDR62, WDR73, WFS1, WNK1, WT1, ZAP70, ZEB2, ZFP57, ZNF423
44	CentoICU™ platinum fast	AARS, AARS2, AASS, ABAT, ABCA12, ABCA3, ABCB11, ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACTA1, ADA, ADAMTS13, ADAR, ADK, ADNP, ADSL, AGK, AGL, AGPAT2, AGRN, AGXT, AHCY, AICDA, AIFM1, AIMP1, AKAP9, AKR1D1, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG14, ALG2, ALG3, ALG6, ALMS1, ALOX12B, ALOXE3, ALPL, ALS2, AMACR, AMT, ANK1, ANKRD26, ANKS6, ANTXR1, AP2S1, AP4B1, AP4E1, AP4M1, AP4S1, APOB, ARG1, ARL6, ARSA, ARSB, ARX, ASL, ASNS, ASPA, ASPM, ASS1, ATP1A3, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATR, ATRX, AUH, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BDNF, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSND, BTD, BTK, C10orf2, C12orf65, C21orf59, CA12, CACNA1C, CACNB2, CALM1, CAMTA1, CASK, CASR, CAST, CAV3, CBS, CCDC103, CCDC114, CCDC78, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDK5RAP2, CDKL5, CDKN1C, CENPJ,

		CEP152, CEP290, CERS3, CFH, CFHR3, CFL2, CFTR, CHAT, CHD7, CHKB, CHM, CHRNA1, CHRNB1, CHRND, CHRNE, CIDEC, CLCNKA, CLCNKB, CLPB, CNTN1, COA5, COL11A1, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COMP, COQ2, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CPS1, CPT1A, CPT2, CR2, CRTAP, CTNS, CTPS1, CTSA, CTSD, CUL4B, CXCR4, CYP11B1, CYP11B2, CYP17A1, CYP4F22, CYP7B1, D2HGDH, DBT, DCLRE1C, DDC, DDOST, DDR2, DECR1, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DIAPH1, DLAT, DLD, DMD, DNA2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC19, DNM2, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM2, DPYD, DRC1, DSP, DST, DUOX2, DUOXA2, DYSF, EDN3, EEF1A2, EGR2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELANE, ENPP1, EPB42, EPCAM, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYA1, EYA4, F10, F11, F13A1, F2, F5, F7, F8, F9, FADD, FAH, FANCA, FANCB, FANCC, FANCD2, FANCL, FARS2, FASTKD2, FBN1, FBP1, FBXL4, FGA, FGB, FGFR2, FGFR3, FGG, FH, FIG4, FKBP14, FKRP, FKTN, FOXC1, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PC2, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GARS, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GFAP, GFM1, GFPT1, GJA1, GJB2, GJB4, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLUD1, GLYCTK, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GP1BA, GP1BB, GP9, GPC3, GPHN, GPSM2, GSS, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HESX1, HEXA, HEXB, HGD, HGF, HIBCH, HLCS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPGD, HRAS, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, ICOS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IGHMBP2, IGLL1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, ISPD, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, IVD, JAG1, JAGN1, JAK3, JAM3, KAT6A, KAT6B, KBTBD13, KCNE1, KCNH1, KCNH2, KCNJ10, KCNJ11, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1B, KLF1, KLHL40, KLHL41, KRAS, KRT5, LAMA2, LAMA3, LAMB3, LAMC2, LAMP2, LAMTOR2, LARS2, LAS1L, LCT, LHX3, LHX4, LIAS, LIG4, LIPA, LIPN, LIPT1, LMBRD1, LMNA, LPIN1, LRBA, LRPPRC, LRRC8A, MAGEL2, MAGT1, MALT1, MAN2B1, MANBA, MAP2K1, MAP2K2, MASTL, MAT1A, MCCC1, MCCC2, MCEE, MCM4, MCPH1, MECP2, MED12, MEF2C, MEGF10, MFN2, MFSD8, MITF, MKKS, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MPC1, MPI, MPL, MPV17, MPZ, MRPL3, MRPL44, MSMO1, MTHFR, MTM1, MTMR14, MTO1, MTR, MTRR, MUSK, MUT, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NDUFV2, NEB, NEFL, NEU1, NEUROG3, NEXN, NFKB2, NFU1, NGF, NHEJ1, NIPAL4, NIPBL, NKX2-1,

		NKX2-5, NLRC4, NLRP3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NR0B1, NR3C2, NRAS, NSD1, NSDHL, NUBPL, OAT, OCLN, OCRL, OPA3, OPHN1, OPLAH, ORC1, ORC4, OTC, OXCT1, PAFAH1B1, PAH, PAX2, PAX3, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH19, PCNT, PDCD10, PDE10A, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX13, PEX19, PEX7, PGAP1, PHGDH, PHOX2B, PIGA, PIGN, PIGT, PIGV, PIK3CD, PKD2, PKHD1, PKLR, PLCB4, PLEC, PLOD1, PLP1, PMM2, PMP22, PNKP, PNP, PNPLA1, PNPO, PNPT1, POGZ, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, PPT1, PRDM16, PRKAG2, PRKDC, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PSAP, PSAT1, PSPH, PTPN11, PTPRC, PTRF, PTRH2, PTS, PURA, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RAC2, RAF1, RAG1, RAG2, RANBP2, RAPSN, RARS2, RB1, RBBP8, RBM8A, RET, RFT1, RFX5, RFX6, RIT1, RMND1, RMRP, RNASEH2C, RNASET2, RNU4ATAC, RORC, RPS19, RRM2B, RYR1, SALL1, SATB2, SBDS, SCN1A, SCN2A, SCN4A, SCN5A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SEPN1, SERAC1, SERPINC1, SERPING1, SFTPB, SFTPC, SFTPD, SHOC2, SIL1, SIX3, SIX5, SKI, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC26A2, SLC26A3, SLC2A1, SLC30A2, SLC33A1, SLC37A4, SLC3A1, SLC46A1, SLC4A1, SLC52A1, SLC52A3, SLC5A1, SLC5A5, SLC6A1, SLC6A3, SLC6A5, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNAI2, SNX10, SOS1, SOX10, SOX2, SOX9, SPAST, SPEG, SPINK5, SPINT2, SPR, SPRED1, SPTA1, SPTAN1, SPTB, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STAT3, STIL, STIM1, STS, STT3B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAT, TAZ, TBC1D24, TBCE, TBX19, TBX5, TCAP, TCN2, TFR2, TG, TGM1, TH, THRA, TJP2, TMCO1, TMEM165, TMEM173, TMEM5, TMEM70, TNFRSF13B, TNFRSF13C, TNFSF4, TNNT1, TP63, TPM2, TPM3, TPO, TPP1, TRIP11, TRMU, TRPV4, TSC1, TSC2, TSFM, TSHB, TSHR, TSPYL1, TTC7A, TTN, TUBA8, TUBB2A, UBA1, UGT1A1, UMPS, UNG, UPB1, UQCRC2, UROD, UROS, WAS, WDPCP, WDR62, WDR73, WFS1, WNK1, WT1, ZAP70, ZEB2, ZFP57, ZNF423
45	CentoMito™ Comprehensive	AARS2, AASS, ABAT, ABCB6, ABCB7, ABCD1, ABCD3, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACSL4, ADCK4, AFG3L2, AGK, AGXT, AIFM1, AK2, ALAS2, ALDH18A1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, AMACR, AMT, APOPT1, ATIC, ATP5A1, ATP5E, ATP7B, ATPAF2, ATXN2, AUH, BAX, BCKDHA, BCKDHB, BCKDK, BCL2, BCS1L, BOLA3, BRIP1, BTD, C10orf2, C12orf65, CA5A, CASP8, CAT, CHCHD10, CISD2, CLPB, CLPP, COA5, COA6, COASY, COMT, COQ2, COQ4, COQ6, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, CPOX, CPS1, CPT1A, CPT1C, CPT2, CRBN, CYB5A, CYB5R3, CYC1,

		CYCS, CYP11A1, CYP11B1, CYP11B2, CYP24A1, CYP27A1, CYP27B1, D2HGDH, DARS2, DBT, DECR1, DGUOK, DHCR24, DHODH, DHTKD1, DIABLO, DLAT, DLD, DMGDH, DMPK, DNA2, DNAJC19, DNAJC3, DNM1L, EARS2, ECHS1, EHHADH, ELAC2, EPHX2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FECH, FH, FKBP10, FOXRED1, FTH1, FXN, GARS, GATM, GCDH, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GLYCTK, GPI, GPT2, GPX1, GRHPR, GSR, GTPBP3, HADH, HADHA, HADHB, HARS2, HAX1, HCCS, HIBCH, HINT1, HK1, HLCS, HMBS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSD17B4, HSD3B2, HSPA9, HSPD1, HTRA2, IARS2, IBA57, IDH2, IDH3B, ISCA2, ISCU, IVD, KARS, KIF1B, KRT5, L2HGDH, LARS2, LIAS, LIPT1, LONP1, LRPPRC, LYRM4, LYRM7, MAOA, MAOB, MARS2, MCCC1, MCCC2, MCEE, MFN2, MGME1, MICU1, MIP, MLH1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS22, MSRB3, MTFMT, MTO1, MTPAP, MTRR, MUT, MUTYH, NADK2, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NNT, NTHL1, NUBPL, OAT, OGDH, OGG1, OPA1, OPA3, OTC, OXCT1, P4HB, PAM16, PANK2, PARK7, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDX1, PET100, PEX11B, PHYH, PINK1, PKLR, PNPLA8, PNPO, PNPT1, POLG, POLG2, PPM1K, PPOX, PRODH, PTGS1, PTRF, PTRH2, PTS, PUS1, PYCR1, PYCR2, QDPR, RARS, RARS2, RDH11, RECQL4, RMND1, RNASEH1, RNASEL, RPIA, RPL35A, RPS14,RRM2B, SARDH, SARS2, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SECISBP2, SERAC1, SFXN4, SLC16A1, SLC19A3, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC37A4, SLC9A6, SNAP29, SOD1, SOD2, SPG7, SPR, SPTLC2, STAR, STOM, SUCLA2, SUCLG1, SUGCT, SUOX, SURF1, TACO1, TARS2, TCIRG1, TIMM44, TIMM8A, TK2, TMEM126A, TMEM70, TMLHE, TPI1, TPK1, TRMU, TRNT1, TSFM, TTC19, TUBB3, TUFM, TXNRD2, TYMP, UNG, UQCC2, UQCRB, UQCRC2, UQCRQ, VARS2, WDR81, WFS1, XPNPEP3, YARS2 + complete coverage of mitochondrial genome
46	CentoMito™ Genome	MT-ND1, MT-ND2, MT-CO1, MT-CO2, MT-ATP8, MT-ATP6, MT-CO3, MT-ND3, MT-ND4L, MT-ND4, MT-ND5, MT-ND6, MT-CYB, MT-TF, MT-RNR1, MT-TV, MT-RNR2, MT-TL1, MT-TI, MT-TQ, MT-TM, MT-TW, MT-TA, MT-TN, MT-TC, MT-TY, MT-TS1, MT-TD, MT-TK, MT-TG, MT-TR, MT-TH, MT-TS2, MT-TL2, MT-TE, MT-TT, MT-TP
47	CentoScreen™ DUO	Analysis of 331 genes for carrier screening

48	CentoScreen™ Paired PACK	Complete panel evaluation of 331 genes for first partner + risk gene analysis for second partner based on the result of first partner
49	CentoScreen™ Paired X-TRA	Risk gene analysis for second partner based on the result of first partner when ordered separately
50	CentoScreen™ SOLO	Analysis of 331 genes for carrier screening
51	Central hypoventilation syndrome panel	RET, GDNF, EDN3, BDNF, ASCL1, PHOX2A, PHOX2B, ZEB2, GFRA1, ECE1, MECP2
52	Cerebellar ataxia panel	COQ8A, APTX, COQ2, COQ9, DNMT1, FXN, PDSS1, PDSS2, POLG, SACS, SETX, SYNE1, TTPA, VLDLR
53	Cerebral cavernous malformations panel	CCM2, KRIT1, PDCD10
54	Ceroid lipofuscinosis panel	ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1
55	Chronic granulomatous disease panel	CYBA, CYBB, NCF1, NCF2, NCF4
56	Ciliary (primary) dyskinesia panel	DNAI1, DNAAF2, DNAAF3, DNAAF5, DNAH5, HYDIN, NME8, DNAH11, DNAI2, RSPH4A, RSPH9, DNAAF1, CCDC39, CCDC40, DNAL1, CCDC103, LRRC6, CCDC114
57	Cleft lip/palate panel	BMP4, IRF6, MSX1, PVRL1, SUMO1, TP63
58	CMT neuropathy panel	AARS, AIFM1, ARHGEF10, BSCL2, COX6A1, DHTKD1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HINT1, HK1, HOXD10, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, PDK3, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SH3TC2, TRIM2, TRPV4, VCP, YARS
59	Coffin-Siris syndrome panel	ARID1A, ARID1B, SMARCA4, SMARCB1, SMARCE1
60	Colon cancer non-polyposis panel	MSH2, MLH1, MSH6, PMS2, EPCAM
61	Colon cancer with polyps panel	APC, BMPR1A, MUTYH, PTEN, SMAD4, STK11
62	Combined pituitary hormone deficiency panel	GHR, HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1

63	Common variable immune deficiency (CVID) panel	ICOS, NFKB2, TNFRSF13B, TNFRSF13C
64	Comprehensive dystonia panel	ADCY5, ANO3, ARSA, ATM, ATP1A3, ATP7B, CACNA1B, CIZ1, COL6A3, DRD2, GCDH, GCH1, GNAL, HPCA, KCNMA1, KCTD17, PANK2, PARK2, PLA2G6, PNKD, PRKRA, PRRT2, RELN, SGCE, SLC2A1, SLC6A3, SPR, TH, THAP1, TIMM8A, TOR1A, TUBB4A
65	Comprehensive epilepsy panel	ACY1, ADSL, ALDH7A1, AMT, ARHGEF15, ARHGEF9, ARX, ASAH1, CACNA1H, CACNB4, CDKL5, CERS1, CHRNA2, CHRNA4, CHRNB2, CLCN2, CNTNAP2, CPA6, CPT2, CSTB, DEPDC5, DRD2, EFHC1, EPM2A, FOLR1, FOXG1, GABRA1, GABRB3, GABRD, GABRG2, GAMT, GCSH, GLDC, GOSR2, GRIN2A, GRIN2B, JRK, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, LGI1, MAPK10, MBD5, MECP2, MEF2C, MFSD8, MTHFR, MTOR, NEDD4L, NEU1, NHLRC1, NOL3, NRXN1, PCDH19, PIGA, PIGO, PIGV, PLCB1, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRICKLE2, PRRT2, QARS, RBFOX1, RBFOX3, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SGCE, SLC13A5, SLC19A3, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A8, SLC9A6, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STXBP1, SYN1, SYNGAP1, SZT2, TBC1D24, TBCE, TCF4, TPP1, TREX1, TSC1, TSC2, UBE3A, WWOX, ZEB2
66	Comprehensive pulmonary disease panel	ABCA3, ACVRL1, AP3B1, ASCL1, BDNF, BLOC1S3, BLOC1S6, BMPR2, CCDC39, CCDC40, CFTR, CSF2RA, CSF2RB, DKC1, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DOCK8, DTNBP1, EDN3, EFEMP2, ELMOD2, ELN, ENG, FBLN5, FBN1, FLCN, FOXF1, GDNF, HPS1, HPS3, HPS4, HPS5, HPS6, LTBP4, MUC5B, NKX2-1, NME8, NOP10, PARN, PHOX2B, RET, RSPH1, RSPH4A, RSPH9, RTEL1, SCNN1A, SCNN1B, SCNN1G, SERPINA1, SFTPA1, SFTPA2, SFTPB, SFTPC, SFTPD, SMAD9, STAT3, TERC, TERT, TINF2, TSC1, TSC2
67	Comprehensive SCID panel	ADA, AK2, CD3D, CD3E, CD247, DCLRE1C, FOXN1, IL2RG, IL7R, JAK3, LIG4, NHEJ1, ORAI1, PNP, PTPRC, RAC2, RAG1, RAG2, RMRP, STAT5B, STIM1, TBX1, ZAP70
68	Cone-rod and cone dystrophy panel	ABCA4, ADAM9, AIPL1, BEST1, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CERKL, CNGB3, CNNM4, CRX, GUCA1A, GUCY2D, KCNV2, PDE6C, PDE6H, PITPNM3, PROM1, PRPH2, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SEMA4A, UNC119
69	Congenital adrenal hyperplasia panel	CYP21A2, CYP17A1, CYP11B1, HSD3B2, POR, STAR

70	Congenital dyserythropoietic anemia panel	C15orf41, CDAN1, GATA1, KLF1, SEC23B
71	Congenital glycosylation disease panel	ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B4GALT1, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPM1, DPM2, DPM3, GMPPA, GNE, LARGE, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SLC35A1, SLC35A2, SLC35C1, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TUSC3
72	Congenital heart defects panel	CFC1, CITED2, CRELD1, FOXH1, GATA4, GATA6, GDF1, NKX2-5, NOTCH1, TBX1, TBX20, ZFPM2
73	Congenital ichthyosis panel	ABCA12, ALOX12B, ALOXE3, CERS3, CYP4F22, LIPN, NIPAL4, PNPLA1, TGM1
74	Congenital myasthenic syndrome panel	AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, GFPT1, LAMB2, MUSK, PLEC, RAPSN, SCN4A
75	Congenital myopathy panel	ACTA1, BIN1, CCDC78, CFL2, CNTN1, DNM2, FHL1, KBTBD13, MAMLD1, MTM1, MTMR14, MYF6, MYH7, NEB, RYR1, SELENON, TNNT1, TPM2, TPM3
76	Congenital neutropenia panel	CSF3R, CXCR4, ELANE, G6PC3, GFI1, HAX1, JAGN1, RAC2, SBDS, VPS45, WAS
78	Congenital sideroblastic anemia panel	ABCB7, ALAS2, GLRX5, PUS1, SLC19A2, SLC25A38, TRNT1, YARS2
79	Cornelia de Lange syndrome panel	AFF4, HDAC8, KMT2A, NIPBL, RAD21, SMC1A, SMC3, TAF6
80	Craniosynostosis and craniofacial disorders panel	EFNB1, ERF, FGFR1, FGFR2, FGFR3, FREM1, GLI3, IFT43, IFT122, IL11RA, MEGF8, MSX2, POR, RAB23, RECQL4, SKI, TCF12, TGFBR1, TGFBR2, TWIST1, WDR19, WDR35
81	Cutis laxa panel	ALDH18A1, ATP6V0A2, ATP7A, EFEMP2, ELN, FBLN5, LTBP4, PYCR1
82	Deafness, non-syndromic sensorineural autosomal dominant panel	ACTG1, CCDC50, COCH, COL11A2, CRYM, DFNA5, DIABLO, DIAPH1, DIAPH3, EYA4, GJB2, GJB3, GJB6, GRHL2, KCNQ4, MIR96, MYH14, MYH9, MYO6, MYO7A, POU3F4, POU4F3, PRPS1, SIX1, SLC17A8, SMPX, TECTA, TJP2, TMC1, WFS1

83	Deafness, non-syndromic sensorineural autosomal recessive panel	CDH23, CLDN14, COL11A2, DFNB31, DFNB59, ESPN, ESRRB, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRXCR1, HGF, ILDR1, KCNJ10, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MSRB3, MYO15A, MYO3A, MYO6, MYO7A, OTOA, OTOF, PCDH15, POU3F4, PRPS1, PTPRQ, RDX, SERPINB6, SLC12A1, SLC26A4, SLC26A5, SMPX, STRC, TECTA, TMC1, TMIE, TMPRSS3, TPRN, TRIOBP, USH1C
84	Dejerine-Sottas syndrome panel	MPZ, PMP22, PRX, EGR2, GJB1
85	Dementia panel	APOE, APP, CHMP2B, CSF1R, FUS, GRN, MAPT, PRNP, PSEN1, PSEN2, SORL1, TARDBP, TREM2, UBE3A, VCP
86	Diabetes neonatal panel	ABCC8, FOXP3, G6PC2, GCK, GLIS3, INS, INSR, KCNJ11, NEUROG3, PDX1
87	Diamond-Blackfan anemia panel	GATA1, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, TSR2
88	Dolichoectasia panel	COL4A1, COL4A2, GAA, MMP3, PKD1, PKD2, SLC2A10
89	Dopa-responsive dystonia panel	GCH1, TH, SPR
90	Dravet syndrome panel	SCN1A, GABRG2, SCN2A, SCN9A

91	Early infantile epileptic encephalopathy panel	AARS, ALG13, ARHGEF9, ARV1, ARX, CACNA1A, CDKL5, DNM1, DOCK7, EEF1A2, FRRS1L, GABRA1, GABRB3, GNAO1, GRIN2B, GUF1, HCN1, ITPA, KCNA2, KCNB1, KCNQ2, KCNT1, NECAP1, PCDH19, PIGA, PLCB1, PNKP, SCN1A, SCN2A, SCN8A, SCN9A, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SPTAN1, ST3GAL3, STXBP1, SZT2, TBC1D24, WWOX
92	Ehlers-Danlos syndrome and related disorders panel	ADAMTS2, ATP7A, B3GALT6, B3GAT3, B4GALT7, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FKBP14, FLNA, GORAB, LTBP4, PLOD1, PRDM5, PYCR1, RIN2, SLC39A13, TNXB, ZNF469
93	Epidermolysis bullosa panel	CHST8, COL17A1, COL7A1, CSTA, DSG1, DSP, DST, EXPH5, FERMT1, ITGA3, ITGA6, ITGB4, JUP, KRT14, KRT5, LAMA3, LAMB3, LAMC2, MMP1, PKP1, PLEC, TGM5
94	Epilepsy (absence) in childhood panel	CACNA1H, GABRA1, GABRB3, GABRG2, JRK, SLC2A1

95	Epilepsy (generalized) with febrile seizures panel	GABRD, GABRG2, SCN1A, SCN1B, SCN2A, SCN9A
96	Epilepsy (partial) hereditary panel	CACNA1H, CACNB4, CHRNA2, CHRNA4, CHRNB2, CLCN2, CPA6, DEPDC5, EFHC1, GABRA1, GABRB3, GABRD, GABRG2, JRK, KCNMA1, KCNQ2, KCNQ3, KCNT1, LGI1, MT-ATP6, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC2A1, SRPX2
97	Epileptic encephalopathy panel	ACY1, ADSL, ALDH7A1, AMT, ARHGEF9, ARX, CDKL5, CNTNAP2, CPT2, FOLR1, FOXG1, GABRG2, GAMT, GCSH, GLDC, GRIN2A, GRIN2B, KCNJ10, KCNQ2, MAPK10, MECP2, MTHFR, NRXN1, PCDH19, PLCB1, PNKP, PNPO, PRRT2, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC19A3, SLC25A22, SLC2A1, SLC9A6, SPTAN1, STXBP1, TBCE, TCF4, TREX1, UBE3A, ZEB2
98	Episodic ataxia panel	CACNA1A, CACNB4, KCNA1, SLC1A3

99	Familial hemiplegic migraine panel	ATP1A2, CACNA1A, SCN1A
100	Familial hypercholesterolemia panel	APOB, GHR, LDLR, PCSK9
101	Familial thoracic aortic aneurysm panel	ACTA2, BGN, CBS, LOX, MAT2A, MFAP5, MYH11, MYLK
102	Fanconi anemia panel	BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4, UBE2T, XRCC2
103	Fatty acid oxidation disorder panel	ACAD9, ACADM, ACADS, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HSD17B10, PPARG, SLC22A5, SLC25A20, TAZ
104	Female infertility panel	BMP15, CYP21A2, FSHR, LHB, LHCGR, TUBB8, ZP1
105	Flecked retina panel	CHM, EFEMP1, PLA2G5, RDH5, RLBP1, RS1, VPS13B
106	Focal Glomerulonephrosis panel	ACTN4, CD2AP, INF2, NPHS1, NPHS2, TRPC6, WT1

107	Frontotemporal dementia panel	CHCHD10, CHMP2B, CSF1R, DCTN1, FUS, GRN, ITM2B, MAPT, PRNP, PSEN1, PSEN2, SIGMAR1, TARDBP, TREM2, TUBA4A, UBQLN2, VCP

108	Gastric cancer panel, targeted	BMPR1A, CDH1, EPCAM, MLH1, MSH2, MSH6, PMS1, PMS2, SMAD4
109	Glaucoma panel	ACVR1, ASB10, BEST1, CANT1, COL18A1, CYP1B1, FOXC1, LMX1B, LOXL1, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, PITX3, SBF2, WDR36
110	Global infertility panel	AR, CATSPER1, CFTR, FSHB, FSHR, HESX1, LHB, LHCGR, NR5A1, POU1F1, SRY
111	Glycogen storage disease panel (advanced)	GYS1, GYS2, G6PC, SLC37A4, GAA, AGL, GBE1, PYGM, PYGL, PFKM, PHKA2, PGAM2, LDHA, ALDOA, ENO3, PHKB, PHKA1, PGM1, GYG1, PRKAG2, PHKG2
112	Glycogen storage disease panel (basic)	G6PC, SLC37A4, AGL, GBE1

113	Hemophagocytic Lymphohistiocytosis panel	PRF1, UNC13D, STX11, STXBP2
114	Hereditary hemorrhagic telangiectasia panel	ACVRL1, ADAM17, ENG, GDF2, PTPN14, RASA1, SMAD4
115	Hermansky-Pudlak syndrome panel	HPS1, AP3B1, HPS3, HPS4, HPS5, HPS6, DTNBP1, BLOC1S3
116	Heterotaxy panel	ACVR2B, CFAP53, CFC1, CRELD1, FOXH1, GDF1, LEFTY2, MMP21, NKX2-5, NODAL, ZIC3
117	Hirschsprung disease panel	ECE1, EDN3, EDNRB, GDNF, KIF1BP, NRG1, NRTN, RET, SOX10, ZEB2
118	Holoprosencephaly panel	CDON, FGF8, GLI2, GLI3, PTCH1, SHH, SIX3, TGIF1, ZIC2
119	Hyperekplexia panel	ARHGEF9, GLRA1, GLRB, GPHN, SLC6A5
120	Hyperinsulinemic hypoglycemia panel	ABCC8, GCK, GLUD1, HADH, INSR, KCNJ11, SLC16A1

121	Hypomagnesemia panel	CLDN16, CLDN19, CNNM2, EGF, FXYD2, KCNA1, SLC12A3, TRPM6

122	Ichthyosis extended panel	ABCA12, ALOX12B, ALOXE3, AP1S1, CERS3, CLDN1, CYP4F22, EBP, ERCC2, ERCC3, FLG, GJB2, GJB3, GJB4, GTF2H5, KRT1, KRT10, KRT2, LIPN, LOR, MPLKIP, NIPAL4, PEX7, PHYH, PNPLA1, POMP, SLC27A4, SNAP29, SPINK5, ST14, STS, SUMF1, TGM1, TGM5
123	Intrahepatic cholestasis panel	ABCB11, ABCB4, ATP8B1, UGT1A1

124	Joubert syndrome panel	AHI1, ARL13B, B9D1, B9D2, C5orf42, CC2D2A, CEP290, CEP41, CSPP1, EXOC8, GLI3, INPP5E, KIF7, MKS1, NEK8, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423

125	Kallmann syndrome and Hypogonadotropic hypogonadism panel	ANOS1, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NSMF, PROK2, PROKR2, SEMA3A, SPRY4, TAC3, TACR3, WDR11
126	Klippel-feil syndrome panel	GDF3, GDF6, MEOX1, MYO18B

127	Leber congenital amaurosis panel	AIPL1, CABP4, CEP290, CRB1, CRX, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, OTX2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1
128	Leber optic atrophy panel	MT-ATP6, MT-CO1, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND4, MT-ND4L, MT-ND5, MT-ND6

129	Leigh syndrome and mitochondrial encephalopathy panel	ACAD9, COQ8A, AIFM1, APTX, ATPAF2, BCS1L, TWNK, NDUFAF6, COQ2, COQ9, COX10, COX15, COX6B1, DARS2, DGUOK, DLAT, DLD, DNM1L, ETFDH, ETHE1, FASTKD2, FH, FOXRED1, GFER, GFM1, LRPPRC, MPV17, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF4, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, NDUFA12, NDUFA9, NDUFAF5, SDHA, PC, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, RARS2, SCO1, SCO2, SDHAF1, SUCLA2, SUCLG1, SURF1, TACO1, TK2, TMEM70, TSFM, TTC19, TUFM, TYMP
130	Leukodystrophy and peroxisome biogenesis disorders panel	ABCD1, AIMP1, ARSA, ASPA, BEST1, CSF1R, CYP27A1, DARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FA2H, FAM126A, GALC, GFAP, GJC2, HEPACAM, HSPD1, MLC1, NDUFV1, NOTCH3, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PLP1, POLR3A, POLR3B, PSAP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SDHA, SLC16A2, SOX10, SUMF1, TREM2, TREX1, HSD17B4, LMNB1, PEX11B, PHYH, SCP2, SDHAF1, TYROBP
131	Lipodystrophy panel	AGPAT2, BSCL2, CAV1, CIDEC, LIPE, LMNA, PIK3R1, PLIN1, PPARG, PTRF
132	Lissencephaly and brain malformation panel	ACTB, ACTG1, ADGRG1, ARX, CDK5, COL6A1, COL6A2, COL6A3, DCX, DYNC1H1, EOMES, FKRP, FKTN, ISPD, KATNB1, KIF2A, KIF5C, LAMA2, LAMB1, LARGE, LMNA, NDE1, PAFAH1B1, POMGNT1, POMT1, POMT2, RELN, SELENON, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, VLDLR, WDR62, YWHAE
133	Long QT syndrome panel	AKAP9, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN
134	Lysosomal storage disease panel	ARSA, FUCA1, GALC, GBA, GLB1, GNPTAB, GUSB, HEXA, HEXB, MAN2B1, MANBA, NAGA, SMPD1

135	Male infertility panel	AR, CATSPER1, CFTR, FSHR, LHCGR
136	Malignant hyperthermia panel	CACNA1S, RYR1
137	Maple syrup urine disease panel	BCKDHA, BCKDHB, DBT, DLD
138	Marfan, Loeys-Dietz syndrome and related disorders panel	COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, NOTCH1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2

139	Meckel syndrome panel	MKS1, TMEM216, TMEM67, CEP290, RPGRIP1L, CC2D2A, NPHP3, TCTN2, B9D1, B9D2, TMEM231
140	Melanoma panel	BAP1, CDK4, CDKN2A, MITF, TERT)
141	Mental retardation, X-linked panel	ABCD1, ACSL4, AFF2, AP1S2, ARHGEF6, ARHGEF9, ARX, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CDKL5, CUL4B, DCX, DKC1, DLG3, ELK1, FANCB, FGD1, FLNA, FMR1, FTSJ1, GDI1, GK, GPC3, GRIA3, HCCS, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IL1RAPL1, KDM5C, KIAA2022, KLF8, L1CAM, LAMP2, MAGT1, MAOA, MBTPS2, MECP2, MED12, MID1, MTM1, NDP, NDUFA1, NHS, NLGN3, NLGN4X, NSDHL, NXF5, OCRL, OFD1, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PLP1, PORCN, PQBP1, PRPS1, RAB39B, RPL10, RPS6KA3, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMC1A, SMS, SOX3, SYN1, SYP, TIMM8A, TSPAN7, UBE2A, UPF3B, ZCCHC12, ZDHHC15, ZDHHC9, ZNF711, ZNF81
142	Metabolic myopathies panel	ABHD5, ACADVL, AGL, CPT2, ENO3, ETFA, ETFB, ETFDH, GAA, GBE1, GYG1, GYS1, LDHA, LPIN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, PRKAG2, PYGM, SLC22A5, SLC25A20, TAZ
143	Metaphyseal dysplasia panel	ANKH, CDKN1C, FLNA, MMP9, MMP13, NKX3-2, RMRP, RUNX2
144	Methylmalonic acidemia panel (advanced)	ABCD4, ACSF3, CD320, LMBRD1, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, SUCLA2, SUCLG1
145	Methylmalonic acidemia panel (basic)	MCEE, MMAA, MMAB, MMADHC, MUT
146	Micro syndrome panel	ALDH18A1, CREBBP, CUL7, RAB18, RAB3GAP1, RAB3GAP2, TBC1D20
147	Microcephaly panel	AKT3, AP4M1, ARFGEF2, ASPM, ASXL3, ATR, ATRX, CASC5, CASK, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CEP135, CEP152, CEP63, CHMP1A, CRIPT, DYRK1A, EFTUD2, IER3IP1, KATNB1, KIF11, MCPH1, MED17, MFSD2A, MSMO1, NDE1, NHEJ1, NIN, ORC1, PCNT, PHC1, PLK4, PNKP, PYCR2, QARS, RBBP8, SASS6, SLC25A19, STAMBP, STIL, TRMT10A, TUBB2B, TUBGCP4, TUBGCP6, WDR62, ZEB2, ZNF335
148	Microphthalmia/anophthalmia/coloboma spectrum panel	ABCB6, ALDH1A3, BCOR, BMP4, CHD7, ERCC1, ERCC2, ERCC5, ERCC6, FOXE3, FOXL2, FRAS1, FREM1, FREM2, GDF3, GDF6, GJA1, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MFRP, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PRSS56, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SALL2, SHH, SIX3, SIX6, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, VAX1, VSX2
149	MODY panel	ABCC8, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, NKX2-2, PAX4, PDX1, RFX6, ZFP57
150	Mucopolysaccharidosis panel	ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, IDS, IDUA, NAGLU, SGSH

151	Multiple endocrine neoplasias /paraganglioma/pheochromocytoma panel	CDKN1B, MAX, MEN1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL
152	Multiple epiphyseal dysplasia panel	COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2
153	Muscular dystrophy panel	ANO5, CAPN3, CAV3, CLCN1, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DYSF, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GMPPB, GNE, HNRNPDL, ISPD, LAMA2, LIMS2, LMNA, MYOT, PLEC, POMGNT1, POMGNT2, POMK, POMT1, POMT2, SELENON, SGCA, SGCB, SGCD, SGCE, SGCG, SMCHD1, SYNE1, SYNE2, TCAP, TNPO3, TRAPPC11, TRIM32, TTN, VCP
154	Muscular dystrophy-dystroglycanopathy type A panel	B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE, POMGNT1, POMGNT2, POMK, POMT1, POMT2, TMEM5
155	Myeloid Tumor Panel	ASXL1, CEBPA, DNMT3A, ETV6, EZH2, IDH1, IDH2, KIT, KRAS, NPM1, NRAS, PTPN11, RAD21, RUNX1, SF3B1, SMC1A, SMC3, STAG2, TET2, TP53, U2AF1, WT1
156	Myoclonic dystonia panel	SGCE, DRD2, TOR1A
157	Myoclonic epilepsy panel	ASAH1, CACNB4, CERS1, CSTB, DRD2, EFHC1, EPM2A, GABRA1, GABRD, GLDC, GOSR2, NEU1, NHLRC1, NOL3, POLG, PRICKLE1, PRICKLE2, SCARB2, SGCE
158	Myofibrillar myopathy panel	BAG3, CRYAB, DES, DNAJB6, FHL1, FLNC, LDB3, MYOT
159	Myopathy-rhabdomyolysis syndrome panel	ACAD9, ACADM, ACADVL, AGL, AMPD1, CPT2, ETFA, ETFB, GAA, GYS1, HADHA, HADHB, LPIN1, OPA1, OPA3, PFKM, PGAM2, PGM1, PHKA1, POLG, POLG2, PYGM, RRM2B, SUCLA2, TK2, TWNK, TYMP

160	Nemaline myopathy panel	ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, NEB, TNNT1, TPM2, TPM3
161	Neonatal mitochondrial hepatopathies panel	BCS1L, DGUOK, FAH, GFM1, HAMP, MPV17, NBAS, POLG, RRM2B, TFR2, TRMU

162	Nephronophthisis panel	NPHP1, INVS, NPHP3, NPHP4, ANKS6, IQCB1, CEP164, CEP290, GLIS2, RPGRIP1L, NEK8, SDCCAG8, ZNF423
163	Nephrotic syndrome panel	ARHGDIA, DGKE, LAMB2, NPHS1, NPHS2, PLCE1, WT1
164	Neurofibromatosis panel	NF1, NF2, SMARCB1, SPRED1
165	Neuronal migration disorders panel	ACTB, ACTG1, ARFGEF2, ARX, COL18A1, COL4A1, CPT2, DCX, EMX2, EOMES, FGFR3, FH, FKRP, FKTN, FLNA, ADGRG1, IER3IP1, ISPD, LAMA2, LAMC3, LARGE, MED12, MEF2C, OCLN, PAFAH1B1, PAX6, PEX7, POMGNT1, POMT1, POMT2, PQBP1, RAB18, RAB3GAP1, RAB3GAP2, RELN, SNAP29, SRPX2, TUBA1A, TUBA8, TUBB2B, TUBB3, VDAC1, WDR62
166	Non ketotic hyperglycinemia panel	AMT, GCSH, GLDC
167	Non-dystrophic myotonia congenita panel	ATP2A1, CACNA1S, CAV3, CLCN1, HINT1, HSPG2, KCNA1, KCNE3, SCN4A
168	Nonsyndromic hypotrichosis panel	APCDD1, CDSN, DSG4, HR, KRT71, KRT74, LIPH, LPAR6, RPL21, SNRPE`
169	Noonan - CFC syndrome panel	BRAF, CBL, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, SPRED1

170	Obesity panel	ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CUL4B, DYRK1B, GNAS, IFT27, LEP, LEPR, LZTFL1, MAGEL2, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, POMC, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B, WDPCP
171	Oculomotor apraxia panel	APTX, PIK3R5, PNKP, SETX
172	Ophthalmoplegia (progressive external) panel	DNA2, OPA1, POLG, POLG2, RRM2B, SLC25A4, TWNK, TYMP
173	Optic atrophy panel	ACO2, AFG3L2, C12ORF65, CISD2, MFN2, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, TIMM8A, TMEM126A, WFS1

174	Osteogenesis imperfecta and low bone density disorders panel	LPL, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, P3H1, LRP5, PLOD2, PLS3, PPIB, SERPINF1, SERPINH1, SP7, TMEM38B, WNT1
175	Osteopetrosis and high bone density disorders panel	ANKH, CA2, CLCN7, COL1A1, GJA1, HPGD, LRP5, MTAP, OSTM1, PLEKHM1, PTDSS1, SLCO2A1, SNX10, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP
176	Ovarian cancer panel, targeted	BARD1, BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MRE11A, MSH2, MSH6, NBN, PMS1, PMS2, RAD50, RAD51C, RAD51D, STK11, TP53

177	Pancreatic cancer panel, targeted	APC, ATM, BMPR1A, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS1, PMS2, PRSS1, SMAD4, STK11
178	Pancreatitis panel	CFTR, CPA1, CTRC, PRSS1, SPINK1
179	Parkinsons disease panel	ATP13A2, ATP1A3, ATP6AP2, DCTN1, DNAJC6, FBXO7, FTL, FUS, GBA, GCH1, GIGYF2, HTRA2, LRRK2, MAPT, PARK2, PARK7, PINK1, PLA2G6, PRKRA, SLC30A10, SLC6A3, SNCA, SNCB, SPR, SYNJ1, TAF1, TH, TMEM230, UCHL1, VPS35
180	Periodic fever syndrome panel	ELANE, LPIN2, MEFV, MVK, NLRP3, PSTPIP1, TNFRSF1A
181	PGL / PCC / GIST panel, targeted	GDNF, KIF1B, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, TP53, VHL
182	Pheochromocytoma panel	MAX, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL
183	Polycystic kidney panel	BICC1, PKD1, PKD2, NOTCH2, PKHD1
184	Pontocerebellar hypoplasia panel	CASK, TSEN2, TSEN34, TSEN54, OPHN1, RARS2, VRK1, EXOSC3, CHMP1A
185	Primary antibody deficiency panel	ADA, AICDA, ATM, BLNK, BTK, CD19, CD40, CD40LG, CD79A, CD79B, CD81, CR2, DCLRE1C, ICOS, IGHM, IGLL1, IKBKG, IL2RG, LRBA, LRRC8A, MS4A1, NCF1, NFKB2, NFKBIA, PIK3CD, PIK3R1, PLCG2, PRKDC, PTPRC, RAG1, RAG2, SH2D1A, TNFRSF13B, TNFRSF13C, UNG, XIAP
186	Prostate cancer panel	BRCA1, BRCA2, CHEK2, HOXB13, MLH1, MSH2, MSH6, NBN, PTEN, TP53

187	Pseudohypoaldosteronism panel	CUL3, HSD11B2, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4
188	Pulmonary hypertension panel	ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, SMAD9

189	Refsum disease panel	PEX1, PEX2, PEX26, PEX7, PHYH
190	Renal cancer panel, targeted	EPCAM, FH, FLCN, HNF1A, HNF1B, MET, MITF, MLH1, MSH2, MSH6, PMS1, PMS2, PTEN, SDHB, SDHD, TSC1, TSC2, VHL, WT1
191	Renal tubular acidosis panel	ATP6V0A4, ATP6V1B1, CA2, EHHADH, HNF4A, SLC34A1, SLC4A1, SLC4A4
192	Retinitis pigmentosa panel, autosomal dominant	ABCA4, BEST1, CA4, CRX, CLRN1, FSCN2, GUCA1B, IMPDH1, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RP2, RP9, RPE65, RPGR, SEMA4A, SNRNP200, TOPORS
193	Retinitis pigmentosa panel, autosomal recessive	ABCA4, ARL6, BBS1, BEST1, C2ORF71, C8ORF37, CERKL, CNGA1, CNGB1, CRB1, DHDDS, EYS, FAM161A, FLVCR1, GNPTG, IDH3B, IMPG2, LRAT, MAK, MERTK, NR2E3, NRL, PDE6A, PDE6B, PDE6G, PRCD, PROM1, RBP3, RDH12, RGR, RHO, RLBP1, RP1, RP2, RPE65, RPGR, SAG, SEMA4A, SPATA7, TTC8, TULP1, USH2A, ZNF513

194	SCA comprehensive panel	ABCB7, ABHD12, ABHD5, ACADVL, AFG3L2, ANO10, APTX, ATCAY, ATM, ATP2B3, CA8, CACNA1A, CCDC88C, COQ8A, CWF19L1, DNMT1, EEF2, ELOVL4, ELOVL5, FGF14, FXN, GRID2, GRM1, ITPR1, KCNC3, KCND3, PDYN, PRKCG, RUBCN, SACS, SETX, SIL1, SLC1A3, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TGM6, TPP1, TTBK2, TTPA, TWNK, VAMP1, WWOX, ZNF592
195	SCA repeat expansion panel	ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP
196	SCA sequencing panel	ABCB7, ABHD12, ABHD5, ACADVL, AFG3L2, ANO10, APTX, ATCAY, ATM, ATP2B3, CA8, CACNA1A, CCDC88C, COQ8A, CWF19L1, DNMT1, EEF2, ELOVL4, ELOVL5, FGF14, FXN, GRID2, GRM1, ITPR1,

		KCNC3, KCND3, PDYN, PRKCG, RUBCN, SACS, SETX, SIL1, SLC1A3, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TGM6, TPP1, TTBK2, TTPA, TWNK, VAMP1, WWOX, ZNF592
197	Seckel syndrome panel	ATR, RBBP8, CENPJ, CEP152, CEP63, ATRIP
198	Skeletal dysplasia ciliopathy panel	DYNC2H1, EVC, EVC2, IFT43, IFT80, IFT122, IFT140, IFT172, NEK1, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60
199	Skeletal dysplasia extended panel	ALPL, ARSE, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, DDR2, EBP, FGFR3, FLNB, HSPG2, INPPL1, LBR, LIFR, MMP13, MMP9, NKX3-2, NSDHL, PEX7, PTH1R, RMRP, SBDS, SLC26A2, SLC35D1, SOX9, TRIP11, TRPV4
200	Skin cancer panel, targeted	CDKN2A, EPCAM, MC1R, MITF, MLH1, MSH2, MSH6, PMS1, PMS2, POT1, PTCH1, XRCC3
201	SMN negative spinal muscular atrophy panel	ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, GARS, HSPB1, HSPB3, HSPB8, IGHMBP2, TRPV4, UBA1, VAPB
202	Solid Tumor Panel	ABL1, AKT1, ALK, APC, AR, ARID1A, ASXL1, ATM, AXL, BRAF, CDH1, CDK4, CDKN2A, CTNNB1, DDR2, EGFR, ERBB2, FGFR1, FGFR2, FGFR3, FGFR4, GNA11, GNAQ, GNAS, HRAS, IDH1, IDH2, JAK2, JAK3, KDM6A, KDR, KIT, KMT2A, KMT2C, KMT2D, KRAS, MAP2K1, MET, MLH1, MTOR, NF1, NOTCH1, NRAS, NTRK3, PDGFRA, PDGFRB, PIK3CA, PIK3R1, PTCH1, PTEN, PTPN11, RB1, RET, ROS1, SMAD4, SMARCA4, SMARCB1, SMO, STK11, TP53, TSC1, TSHR, VHL
203	Spastic paraplegia panel complete	ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ARSI, ATL1, ATP2B4, B4GALNT1, BICD2, BSCL2, C12ORF65, C19orf12, CCT5, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FLRT1, GBA2, GJC2, HSPD1, KIAA0196, KIF1A, KIF1C, KIF5A, L1CAM, MAG, MARS, NIPA1, NT5C2, PGAP1, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RTN2, SACS, SLC16A2, SLC33A1, SPAST, SPG11, SPG20, SPG21, SPG7, TECPR2, TFG, TTR, USP8, VAMP1, VPS37A, WDR48, ZFR, ZFYVE26, ZFYVE27
204	Spastic paraplegia panel, autosomal dominant	ALDH18A1, ATL1, ATP2B4, BICD2, BSCL2, HSPD1, KIAA0196, KIF5A, NIPA1, REEP1, RTN2, SLC33A1, SPAST, TTR, VAMP1, ZFYVE27
205	Spastic paraplegia panel, autosomal recessive	ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ARSI, B4GALNT1, C12ORF65, C19orf12, CCT5, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FLRT1, GBA2, GJC2, KIF1A, KIF1C, L1CAM, MAG, NT5C2, PLP1, PNPLA6, REEP2, SACS, SLC16A2, SPG11, SPG20, SPG21, SPG7, TECPR2, TFG, USP8, VPS37A, WDR48, ZFYVE26
206	Spherocytosis panel	ANK1, EPB42, SLC4A1, SPTA1, SPTB

207	Stargardt disease panel	ABCA4, BEST1, C1QTNF5, CDH3, CNGB3, ELOVL4, FSCN2, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3
208	Stickler syndrome panel	COL2A1, COL9A1, COL9A2, COL11A1, COL11A2
209	Surfactant metabolism dysfunction panel	ABCA3, CSF2RA, CSF2RB, SFTPA1, SFTPB, SFTPC, SFTPD
210	Susceptibility to atypical mycobacterium disease panel	CYBB, IFNGR1, IFNGR2, IKBKG, IL12A, IL12B, IL12RB1, IL12RB2, IRF8, ISG15, STAT1, TYK2
211	Syndromic autism panel	ADNP, ADSL, ALDH5A1, AMT, ANKRD11, ARID1B, BRAF, CACNA1C, CDKL5, CHD2, CHD7, CNTNAP2, CREBBP, DHCR7, EHMT1, FOXG1, FOXP1, GRIP1, HDAC8, HOXA1, HPRT1, MAGEL2, MECP2, MED12, MID1, NHS, NIPBL, NRXN1, NSD1, PCDH19, POGZ, PQBP1, PTEN, PTPN11, RAD21, RAI1, SCN1A, SCN2A, SETD2, SLC6A1, SLC6A8, SMC1A, SMC3, TBL1XR1, TCF4, TSC1, TSC2, UBE3A, VPS13B, ZEB2

212	Thrombocytopenia panel	ADAMTS13, ANKRD26, CYCS, GATA1, GP1BA, GP1BB, GP9, ITGA2B, ITGB3, MASTL, MPL, MYH9, RUNX1, WAS
213	Thyroid cancer panel, targeted	APC, PTEN, RET
214	Tuberous sclerosis panel	TSC1, TSC2

215	Ullrich muscular dystrophy panel	COL6A1, COL6A2, COL6A3
216	Urea cycle disorder panel	ARG1, ASL, ASS1, CPS1, NAGS, OTC
217	Usher syndrome panel	CDH23, CIB2, CLRN1, DFNB31, ADGRV1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A
218	Uterine cancer panel, targeted	Uterine cancer panel, targeted (EPCAM, MLH1, MSH2, MSH6, PMS1, PMS2, PTEN

219	Vitreoretinopathy and Wagner syndrome panel	COL2A1, FZD4, LRP5, NDP, TSPAN12, VCAN

220	Waardenburg syndrome panel	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10, TYR

221	Zellweger syndrome panel	PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26

SINGLE GENE ANALYSIS

Sr No.	Single Gene Analysis
1.	17-beta hydroxysteroid dehydrogenase X deficiency (HSD17B10)
2.	17-hydroxylation activity deficiency (CYP17A1)
3.	2-aminoadipic 2-oxoadipic aciduria (DHTKD1)
4.	2-methylbutyrylglycinuria (ACADSB)
5.	3-beta-hydroxysteroid dehydrogenase deficiency type 2 (HSD3B2)
6.	3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCL)
7.	3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency (HMGCS2)
8.	3-hydroxyisobutryl-CoA hydrolase deficiency (HIBCH)
9.	3-methylglutaconic aciduria type 1 (AUH)
10.	3-methylglutaconic aciduria type 3 (OPA3)
11.	3-methylglutaconic aciduria type 5 (DNAJC19)
12.	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome (SERAC1)
13.	3MC syndrome type 1 (MASP1)
14.	3MC syndrome type 2 (COLEC11)
15.	46,XX sex reversal type 1 (SRY)
16.	46,XY gonadal dysgenesis, partial, with minifascicular neuropathy (DHH)
17.	46,XY sex reversal type 8, modifier of (AKR1C4)
18.	5-oxoprolinase deficiency (OPLAH)
19.	6q24-related transient neonatal diabetes mellitus type 1 (UPD chr. 6)
20.	Abetalipoproteinemia (MTTP)
21.	Accelerated tumor formation, susceptibility to (MDM2)
22.	Acetycholinesterase deficiency (ACHE)
23.	Acetyl-CoA carboxylase deficiency (NGS Sequencing: ACACA)
24.	Achalasia addisonianism alacrimia syndrome (AAAS)
25.	Achondrogenesis type 1A (TRIP11)
26.	Achondrogenesis type 1B (SLC26A2)
27.	Achondrogenesis type 2 (COL2A1)
28.	Achondroplasia (FGFR3)
29.	Achromatopsia type 2 (CNGA3)
30.	Achromatopsia type 3 (CNGB3)

31.	Achromatopsia type 4 (GNAT2)
32.	Achromatopsia type 6 (PDE6H)
33.	Acne inversa familial type 3 (PSEN1)
34.	Acrocallosal syndrome (KIF7)
35.	Acrodermatitis enteropathica (SLC39A4)
36.	Acrodysostosis 2 (PDE4D)
37.	Acrodysostosis type 1, with or without hormone resistance (PRKAR1A)
38.	Acrofacial dysostosis 1, Nager type (SF3B4)
39.	Acromegaly, predisposition to, due to germline GPR101 mutation (GPR101)
40.	Acromelic frontonasal dysostosis (ZSWIM6)
41.	Acromesomelic dysplasia, Maroteaux type (NPR2)
42.	Acromicric dysplasia (NGS Sequencing: FBN1)
43.	Acute myeloid leukemia, somatic, DNMT3A related (DNMT3A)
44.	Acyl-CoA medium-chain dehydrogenase deficiency (ACADM)
45.	Acyl-CoA multiple dehydrogenase deficiency (ETFA)
46.	Acyl-CoA multiple dehydrogenase deficiency (ETFB)
47.	Acyl-CoA peroxisomal oxidase deficiency (ACOX1)
48.	Acyl-CoA short-chain dehydrogenase deficiency (ACADS)
49.	Acyl-CoA very long-chain dehydrogenase deficiency (ACADVL)
50.	Adams-Oliver syndrome type 1 (ARHGAP31)
51.	Adams-Oliver syndrome type 2 (DOCK6)
52.	Adams-Oliver syndrome type 3 (RBPJ)
53.	Adams-Oliver syndrome type 4 (EOGT)
54.	Adams-Oliver syndrome type 6 (DLL4)
55.	Adenine phosphoribosyltransferase deficiency (APRT)
56.	Adenocarcinoma of lung, somatic (BRAF)
57.	Adenocarcinoma of lung, somatic (ERBB2)
58.	Adenoma, periampullary, somatic (APC)
59.	Adenosine triphosphate, elevated, of erythrocytes (PKLR)
60.	Adenylosuccinase deficiency (ADSL)
61.	Adrenal adenoma, somatic (MEN1)
62.	Adrenal hyperplasia due to 21-hydroxylase deficiency (CYP21A2)

63.	Adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency (POR)
64.	Adrenal hyperplasia due to steroid 11-beta-hydroxylase deficiency (CYP11B1)
65.	Adrenal hypoplasia (NR0B1)
66.	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete (CYP11A1)
67.	Adrenocorticotropic hormone deficiency (TBX19)
68.	Adrenoleukodystrophy, x-linked (ABCD1)
69.	Adrenoleukodystrophy, x-linked (PLXNB3)
70.	Adrenoleukodystrophy/Adrenomyeloneuropathy (ABCD1)
71.	ADULT syndrome, split hand-foot malformation (TP63)
72.	Afibrinogenemia, congenital (FGA)
73.	Afibrinogenemia, congenital (FGG)
74.	Afibrinogenemia, congenital (FGB)
75.	Agammaglobulinemia and isolated hormone deficiency (BTK)
76.	Agammaglobulinemia type 1, autosomal recessive (IGHM)
77.	Agammaglobulinemia type 1, X-linked (BTK)
78.	Agammaglobulinemia type 2, autosomal recessive (IGLL1)
79.	Agammaglobulinemia type 3, autosomal recessive (CD79A)
80.	Agammaglobulinemia type 4, autosomal recessive (BLNK)
81.	Agammaglobulinemia type 5, autosomal recessive (LRRC8A)
82.	Agammaglobulinemia type 6, autosomal recessive (CD79B)
83.	Agammaglobulinemia type 7, autosomal recessive (PIK3R1)
84.	Agenesis of the corpus callosum with peripheral neuropathy (SLC12A6)
85.	Aicardi-Goutieres syndrome type 1 (TREX1)
86.	Aicardi-Goutieres syndrome type 2 (RNASEH2B)
87.	Aicardi-Goutieres syndrome type 3 (RNASEH2C)
88.	Aicardi-Goutieres syndrome type 4 (RNASEH2A)
89.	Aicardi-Goutieres syndrome type 5 (SAMHD1)
90.	Aicardi-Goutieres syndrome type 6 (ADAR)
91.	Aicardi-Goutieres syndrome type 7 (IFIH1)
92.	Al-Raqad syndrome (DCPS)
93.	Alacrima, achalasia and mental retardation syndrome (GMPPA)
94.	Alagille syndrome type 1 (JAG1)

95.	Alagille syndrome type 2 (NOTCH2)
96.	Aland Island eye disease (CACNA1F)
97.	Alazami syndrome (LARP7)
98.	Albinism, ocular type I, Nettleship-Falls type (GPR143)
99.	Albinism, oculocutaneous nonsyndromic (SLC24A5)
100.	Albinism, oculocutaneous type 1A (TYR)
101.	Albinism, oculocutaneous type 1B (TYR)
102.	Albinism, oculocutaneous type 2 (OCA2)
103.	Albinism, oculocutaneous type 3 (TYRP1)
104.	Albinism, oculocutaneous type 4 (SLC45A2)
105.	Albinism, oculocutaneous type 5 (C10ORF11)
106.	Alexander disease (GFAP)
107.	Alkaptonuria (HGD)
108.	Allan-Herndon-Dudley syndrome (SLC16A2)
109.	Alopecia universalis (HR)
110.	Alpha-2-macroglobulin deficiency (A2M)
111.	Alpha-ketoglutarate dehydrogenase deficiency (OGDH)
112.	Alpha-methylacyl CoA racemase deficiency (AMACR)
113.	Alpha-thalassemia/mental retardation syndrome (ATRX)
114.	Alport syndrome, autosomal recessive (COL4A4)
115.	Alport syndrome, autosomal recessive (COL4A3)
116.	Alport syndrome, X-Linked (COL4A5)
117.	Alstrom syndrome (ALMS1)
118.	Alternating hemiplegia of childhood type 1 (ATP1A2)
119.	Alternating hemiplegia of childhood type 2 (ATP1A3)
120.	Alveolar capillary dysplasia with misalignment of pulmonary veins (FOXF1)
121.	Alzheimer disease type 1 (APP)
122.	Alzheimer disease type 2 (APOE)
123.	Alzheimer disease type 3 (PSEN1)
124.	Alzheimer disease type 4 (PSEN2)
125.	Alzheimers disease, early onset, autosomal dominant (SORL1)
126.	Alzheimers disease, RTN3 related (RTN3)

127.	Amelogenesis imperfecta type 1A (LAMB3)
128.	Amelogenesis imperfecta type 1B (ENAM)
129.	Amelogenesis imperfecta type 1C (ENAM)
130.	Amelogenesis imperfecta type 1E (AMELX)
131.	Amelogenesis imperfecta type 1F (AMBN)
132.	Amelogenesis imperfecta type 1G (FAM20A)
133.	Amelogenesis imperfecta type 1H (ITGB6)
134.	Amelogenesis imperfecta type 2A1 (KLK4)
135.	Amelogenesis imperfecta type 2A2 (MMP20)
136.	Amelogenesis imperfecta type 2A3 (WDR72)
137.	Amelogenesis imperfecta type 2A4 (C4orf26)
138.	Amelogenesis imperfecta type 2A5 (SLC24A4)
139.	Amelogenesis imperfecta type 3 (FAM83H)
140.	Amelogenesis imperfecta type 4 (DLX3)
141.	Amelotin deficiency (AMTN)
142.	Aminoacylase deficiency (ACY1)
143.	Amish infantile epilepsy syndrome (ST3GAL5)
144.	AMP deaminase deficiency, erythrocytic (AMPD3)
145.	Amyloidosis (TTR)
146.	Amyloidosis, familial visceral (APOA1)
147.	Amyloidosis, finnish type (GSN)
148.	Amyloidosis, primary localized cutaneous, type 1 (OSMR)
149.	Amyloidosis, primary localized cutaneous, type 2 (IL31RA)
150.	Amyotrophic lateral sclerosis risk factor (CHGB)
151.	Amyotrophic lateral sclerosis type 1 (SOD1)
152.	Amyotrophic lateral sclerosis type 10 (TARDBP)
153.	Amyotrophic lateral sclerosis type 11 (FIG4)
154.	Amyotrophic lateral sclerosis type 12 (OPTN)
155.	Amyotrophic lateral sclerosis type 14 (VCP)
156.	Amyotrophic lateral sclerosis type 16 (SIGMAR1)
157.	Amyotrophic lateral sclerosis type 17 (CHMP2B)
158.	Amyotrophic lateral sclerosis type 18 (PFN1)

159.	Amyotrophic lateral sclerosis type 2, juvenile (ALS2)
160.	Amyotrophic lateral sclerosis type 21 (MATR3)
161.	Amyotrophic lateral sclerosis type 4 (SETX)
162.	Amyotrophic lateral sclerosis type 6 (FUS)
163.	Amyotrophic lateral sclerosis type 8 (VAPB)
164.	Amyotrophic lateral sclerosis type 9 (ANG)
165.	Amyotrophic lateral sclerosis with frontotemporal dementia (C9orf72)
166.	Amyotrophic lateral sclerosis, CREST related (SS18L1)
167.	Amyotrophic lateral sclerosis, susceptibility to (NEFH)
168.	Amyotrophic lateral sclerosis, VPS54 related (VPS54)
169.	Amyotrophic lateral sclerosis, x-linked juvenile and adult-onset ALS (UBQLN2)
170.	Amyotrophy hereditary neuralgic (SEPT9)
171.	Andersen disease (GBE1)
172.	Androgen insensitivity (AR)
173.	Androgen insensitivity, partial, with or without breast cancer (AR)
174.	Androgen-binding protein deficiency (SHBG)
175.	Anemia dyserythropoietic type 1A (CDAN1)
176.	Anemia dyserythropoietic type 2 (SEC23B)
177.	Anemia, neonatal hemolytic, fatal and near-fatal (SPTB)
178.	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive (SLC25A38)
179.	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive (GLRX5)
180.	Anemia, sideroblastic, with ataxia (ABCB7)
181.	Anemia, sideroblastic, X-linked (ALAS2)
182.	Anemia, X-linked (GATA1)
183.	Angelman syndrome (UBE3A)
184.	Angelman syndrome (chr. 15q11)
185.	Angelman-like syndrome (CDKL5)
186.	Angelman-like syndrome (MECP2)
187.	Angioedema, hereditary (SERPING1)
188.	Angiofibroma, somatic (MEN1)
189.	Anhaptoglobinemia (HP)
190.	Aniridia (PAX6)

191.	Ankyloblepharon-ectodermal defects-cleft lip/palate (TP63)
192.	Anterior segment mesenchymal dysgenesis (PITX3)
193.	Antithrombin III deficiency (SERPINC1)
194.	Antitrypsin-alpha-1 deficiency (SERPINA1)
195.	Antley-Bixler syndrome (FGFR2)
196.	Aortic aneurysm, familial thoracic type 3 (TGFBR2)
197.	Aortic aneurysm, familial thoracic type 4 (MYH11)
198.	Aortic aneurysm, familial thoracic type 5 (TGFBR1)
199.	Aortic aneurysm, familial thoracic type 6 (ACTA2)
200.	Aortic aneurysm, familial thoracic type 7 (MYLK)
201.	Aortic aneurysm, familial thoracic type 8 (PRKG1)
202.	Aortic aneurysm, familial thoracic, MAT2A related (MAT2A)
203.	Aortic valve disease type 1 (NOTCH1)
204.	Aortic valve disease type 2 (SMAD6)
205.	Apert syndrome (FGFR2)
206.	Aplastic anemia (PRF1)
207.	Aplastic anemia (TERC)
208.	Aplastic anemia, SBDS related (SBDS)
209.	Apolipoprotein C-II deficiency (APOC2)
210.	Apparent mineralocorticoid excess (HSD11B2)
211.	Arginase deficiency (ARG1)
212.	Arginine-glycine amidinotransferase deficiency (GATM)
213.	Argininosuccinic aciduria (ASL)
214.	Aromatase deficiency (CYP19A1)
215.	Aromatic L-amino acid decarboxylase deficiency (DDC)
216.	Arrhythmogenic right ventricular cardiomyopathy type 1 (TGFB3)
217.	Arrhythmogenic right ventricular cardiomyopathy type 10 (DSG2)
218.	Arrhythmogenic right ventricular cardiomyopathy type 11 (DSC2)
219.	Arrhythmogenic right ventricular cardiomyopathy type 12 (JUP)
220.	Arrhythmogenic right ventricular cardiomyopathy type 5 (TMEM43)
221.	Arrhythmogenic right ventricular cardiomyopathy type 8 (DSP)
222.	Arrhythmogenic right ventricular cardiomyopathy type 9 (PKP2)

223.	Arrhythmogenic right ventricular dysplasia type 2 (NGS Sequencing: RYR2)
224.	Arterial calcification type 1, generalized, infantile (ENPP1)
225.	Arterial calcification type 2, generalized, infantile (ABCC6)
226.	Arterial Tortuosity Syndrome (SLC2A10)
227.	Arthrogryposis, distal, type 1A (TPM2)
228.	Arthrogryposis, distal, type 1B (MYBPC1)
229.	Arthrogryposis, distal, type 2A (MYH3)
230.	Arthrogryposis, distal, type 2B (MYH3)
231.	Arthrogryposis, distal, type 2B (TNNI2)
232.	Arthrogryposis, distal, type 2B (TNNT3)
233.	Arthrogryposis, distal, type 3 (PIEZO2)
234.	Arthrogryposis, distal, type 5 (PIEZO2)
235.	Arthrogryposis, distal, type 5D (ECEL1)
236.	Arthrogryposis, distal, type 7 (MYH8)
237.	Arthrogryposis, mental retardation, and seizures (SLC35A3)
238.	Arthrogryposis, renal dysfunction, and cholestasis type 1 (VPS33B)
239.	Arthrogryposis, renal dysfunction, and cholestasis type 2 (VIPAS39)
240.	Arthropathy, progressive pseudorheumatoid, of childhood (WISP3)
241.	Arts syndrome (PRPS1)
242.	Asparaginesynthetase deficiency (ASNS)
243.	Aspartylglucosaminuria (AGA)
244.	Asperger syndrome susceptibility X-linked type 2 (NLGN3)
245.	Ataxia and muscle hypotonia (COX20)
246.	Ataxia telangiectasia like disorder (MRE11A)
247.	Ataxia, posterior column, with retinitis pigmentosa (FLVCR1)
248.	Ataxia, progressive seizures, mental deterioration, and hearing loss, MT-TV related (NGS Sequencing: MT-TV)
249.	Ataxia-oculomotor apraxia type 1 (APTX)
250.	Ataxia-oculomotor apraxia type 2 (SETX)
251.	Ataxia-oculomotor apraxia type 3 (PIK3R5)
252.	Ataxia-oculomotor apraxia type 4 (PNKP)
253.	Ataxia-telangiectasia (NGS Sequencing: ATM)
254.	Atelosteogenesis type 1 (FLNB)

255.	Atelosteogenesis type 3 (FLNB)
256.	Athabaskan brainstem dysgenesis syndrome (HOXA1)
257.	Atherosclerosis, SOAT1 related (SOAT1)
258.	Atrial fibrillation type 10 (SCN5A)
259.	Atrial fibrillation type 11 (GJA5)
260.	Atrial fibrillation type 12 (ABCC9)
261.	Atrial fibrillation type 3 (KCNQ1)
262.	Atrial fibrillation type 4 (KCNE2)
263.	Atrial fibrillation type 6 (NPPA)
264.	Atrial fibrillation type 7 (KCNA5)
265.	Atrial septal defect type 2 (GATA4)
266.	Atrial septal defect type 3 (MYH6)
267.	Atrial septal defect type 4 (TBX20)
268.	Atrial septal defect type 5 (ACTC1)
269.	Atrial septal defect type 8 (CITED2)
270.	Atrial septal defect type 9 (GATA6)
271.	Atrial septal defect with atrioventricular conduction defects (NKX2-5)
272.	Atrichia with papular lesions (HR)
273.	Atrioventricular septal defect type 4 (GATA4)
274.	Atrioventricular septal defect type 5 (GATA6)
275.	Atrioventricular septal defect, partial with heterotaxy syndrome (CRELD1)
276.	Attention deficit-hyperactivity disorder (DRD5)
277.	Attention deficit-hyperactivity disorder (DRD4)
278.	Atypical Mycobacterial infection (IL12RB1)
279.	Atypical Mycobacterial infection (IFNGR2)
280.	Atypical Mycobacterial infection (IKBKG)
281.	Atypical Mycobacterial infection (STAT1)
282.	Atypical Mycobacterial infection, IL12RB2 related (IL12RB2)
283.	Auditory neuropathy, autosomal dominant (DIAPH3)
284.	Auriculocondylar syndrome type 1 (GNAI3)
285.	Auriculocondylar syndrome type 2 (PLCB4)
286.	Autism spectrum disorder (BPIFA3)

287.	Autism spectrum disorder (AHNAK2)
288.	Autism spectrum disorder (EN2)
289.	Autism spectrum disorder (RABGGTA)
290.	Autism spectrum disorder (ANKS3)
291.	Autism spectrum disorder, MYO16 related (MYO16)
292.	Autism spectrum, MXRA5 related (MXRA5)
293.	Autism spectrum/ hyperactivity/ bipolar disorder, GRM7 related (GRM7)
294.	Autism susceptibility, X-linked type 1 (NLGN3)
295.	Autism susceptibility, X-linked type 17 (SHANK2)
296.	Autism susceptibility, X-linked type 2 (NLGN4X)
297.	Autism susceptibility, X-linked type 3 (MECP2)
298.	Autism susceptibility, X-linked type 4 (PTCHD1)
299.	Autism susceptibility, X-linked type 5 (RPL10)
300.	Autism, ATP1B4 related (ATP1B4)
301.	Autism, AVPR1A related (AVPR1A)
302.	Autism, C7orf43 related (C7orf43)
303.	Autism, CELF6 related (CELF6)
304.	Autism, EFCAB13 related (EFCAB13)
305.	Autism, FAAH2 related (FAAH2)
306.	Autism, FCRL6 related (FCRL6)
307.	Autism, GYG2 related (GYG2)
308.	Autism, IQCE related (IQCE)
309.	Autism, MBD1 related (MBD1)
310.	Autism, NTNG1 related (NTNG1)
311.	Autism, OR13H1 related (OR13H1)
312.	Autism, OXTR related (OXTR)
313.	Autism, PKHD1L1 related (NGS Sequencing: PKHD1L1)
314.	Autism, RNF128 related (RNF128)
315.	Autism, RRM1 related (RRM1)
316.	Autism, SETD2 related (SETD2)
317.	Autism, SLC22A9 related (SLC22A9)
318.	Autism, UNC13B related (UNC13B)

319.	Autism, ZNF778 related (ZNF778)
320.	Autism/Mental retardation/Angelman syndrome, susceptibility to, ATP10A related (ATP10A)
321.	Autoimmune lymphoproliferative syndrome type 1A (FAS)
322.	Autoimmune lymphoproliferative syndrome type 1B (FASLG)
323.	Autoimmune lymphoproliferative syndrome type 2A (CASP10)
324.	Autoimmune lymphoproliferative syndrome type 2B (CASP8)
325.	Autoimmune lymphoproliferative syndrome type 3 (PRKCD)
326.	Autoimmune polyendocrinopathy syndrome type 1 (AIRE)
327.	Autoinflammation, lipodystroph and dermatosis syndrome (PSMB8)
328.	Avascular necrosis of the femoral head, primary (COL2A1)
329.	Axenfeld-Rieger syndrome type 1 (PITX2)
330.	Axenfeld-Rieger syndrome type 3 (FOXC1)
331.	Azoospermia induced by Y chromosome microdeletions (AZF region)
332.	B-cell expansion with NFKB and T-cell anergy (CARD11)
333.	Bainbridge-Ropers syndrome (ASXL3)
334.	Baller-Gerold syndrome (RECQL4)
335.	Band-like calcification with simplified gyration and polymicrogyria (OCLN)
336.	Baraitser-Winter syndrome type 1 (ACTB)
337.	Baraitser-Winter syndrome type 2 (ACTG1)
338.	Bardet-Biedl syndrome type 1 (BBS1)
339.	Bardet-Biedl syndrome type 10 (BBS10)
340.	Bardet-Biedl syndrome type 11 (TRIM32)
341.	Bardet-Biedl syndrome type 12 (BBS12)
342.	Bardet-Biedl syndrome type 13 (MKS1)
343.	Bardet-Biedl syndrome type 14 (CEP290)
344.	Bardet-Biedl syndrome type 15 (WDPCP)
345.	Bardet-Biedl syndrome type 2 (BBS2)
346.	Bardet-Biedl syndrome type 3 (ARL6)
347.	Bardet-Biedl syndrome type 4 (BBS4)
348.	Bardet-Biedl syndrome type 5 (BBS5)
349.	Bardet-Biedl syndrome type 6 (MKKS)
350.	Bardet-Biedl syndrome type 7 (BBS7)

351.	Bardet-Biedl syndrome type 8 (TTC8)
352.	Bardet-Biedl syndrome type 9 (BBS9)
353.	Bardet-Biedl syndrome, LZTFL1 related (LZTFL1)
354.	Bardet-Biedl syndrome, modifier of, CCDC28B related (CCDC28B)
355.	Bare lymphocyte syndrome, type 2 (RFXANK)
356.	Bare lymphocyte syndrome, type 2, complementation group A (CIITA)
357.	Barth syndrome (TAZ)
358.	Bartter syndrome (SLC12A7)
359.	Bartter syndrome (SLC12A5)
360.	Bartter syndrome (SLC12A3)
361.	Bartter syndrome (SLC12A2)
362.	Bartter syndrome type 1 (SLC12A1)
363.	Bartter syndrome type 2 (KCNJ1)
364.	Bartter syndrome type 3 (CLCNKB)
365.	Bartter syndrome type 4a (BSND)
366.	Bartter syndrome type 4b (CLCNKA)
367.	Basal cell carcinoma type 7, susceptibility to, somatic (TP53)
368.	Basal cell nevus syndrome (PTCH1)
369.	Basal cell nevus syndrome (SUFU)
370.	Basal cell nevus syndrome due to germline PTCH2 mutation (PTCH2)
371.	Basal ganglia calcification type 1, ideopathic (SLC20A2)
372.	Basal ganglia calcification type 4 (PDGFRB)
373.	Basal ganglia calcification type 5, idiopathic (PDGFB)
374.	Basal ganglia calcification type 6, idiopathic (XPR1)
375.	Beare-Stevenson cutis gyrata syndrome (FGFR2)
376.	Beckwith-Wiedemann syndrome (H19)
377.	Beckwith-Wiedemann syndrome (KCNQ1OT1)
378.	Beckwith-Wiedemann syndrome (chr. 11p15)
379.	Beckwith-Wiedemann syndrome (NSD1)
380.	Beckwith-Wiedemann syndrome (CDKN1C)
381.	Bent bone dysplasia syndrome (FGFR2)
382.	Bernard Soulier syndrome type A1 (GP1BA)

383.	Bernard Soulier syndrome type A2 (GP1BA)
384.	Bernard Soulier syndrome type B (GP1BB)
385.	Bernard Soulier syndrome type C (GP9)
386.	Bestrophinopathy (BEST1)
387.	Beta-Galactosamide alpha-2,6-Sialyltransferase 2 deficiency (ST6GAL2)
388.	Beta-ureidopropionase deficiency (UPB1)
389.	Bethlem myopathy (COL6A2)
390.	Bethlem myopathy (COL6A1)
391.	Bethlem myopathy type 1 (COL6A3)
392.	Bethlem myopathy type 2 (NGS Sequencing: COL12A1)
393.	Bicuspid aortic valve (TIMP1)
394.	Bietti crystalline corneoretinal dystrophy (CYP4V2)
395.	Bifid nose (FREM1)
396.	Bile acid malabsorption, primary (SLC10A2)
397.	Bile acid synthesis defect type 2, congenital (AKR1D1)
398.	Bile acid synthesis defect type 3, congenital (CYP7B1)
399.	Bile acid synthesis defect type 4, congenital (AMACR)
400.	Biotinidase deficiency (BTD)
401.	Birt-Hogg-Dube syndrome (FLCN)
402.	Bjornstad syndrome (BCS1L)
403.	Bladder cancer, HRAS related, somatic (HRAS)
404.	Bladder cancer, somatic (FGFR3)
405.	Bladder cancer, somatic (KRAS)
406.	Bladder cancer, TSC1-related, somatic (TSC1)
407.	Blau syndrome (NOD2)
408.	Bleeding disorder, platelet-type 15 (ACTN1)
409.	Bleeding disorder, platelet-type 17 (GFI1B)
410.	Bleeding disorder, platelet-type 8 (P2RY12)
411.	Blepharophimosis, epicanthus inversus, and ptosis (FOXL2)
412.	Blepharophimosis-ptosis-intellectual disability syndrome (UBE3B)
413.	Bloom syndrome (BLM)
414.	Bohring-Opitz syndrome (ASXL1)

415.	Bone marrow failure syndrome type 1 (SRP72)
416.	Bone marrow failure syndrome type 2 (ERCC6L2)
417.	Bone mineral density QTL18, osteoporosis (PLS3)
418.	Borjeson-Forssman-Lehmann syndrome (PHF6)
419.	Bothnia retinal dystrophy (RLBP1)
420.	Brachydactyly type A1C (GDF5)
421.	Brachydactyly type A2 (BMP2)
422.	Brachydactyly type A2 (BMPR1B)
423.	Brachydactyly type B1 (ROR2)
424.	Brachydactyly type E1 (HOXD13)
425.	Brachydactyly-mental retardation syndrome (HDAC4)
426.	Brachydactyly-syndactyly syndrome (HOXD13)
427.	Bradyopsia (RGS9BP)
428.	Bradyopsia (RGS9)
429.	BRAF somatic Hotspot: c.1799T>A p.V600E (BRAF)
430.	BRAF, selective sequencing of exon 15 (BRAF)
431.	Branched-chain aminotransferase 1 deficiency (BCAT1)
432.	Branched-chain aminotransferase 2 deficiency (BCAT2)
433.	Branched-chain ketoacid dehydrogenase kinase deficiency (BCKDK)
434.	Branchiooculofacial syndrome (TFAP2A)
435.	Branchiootic syndrome type 1 (EYA1)
436.	Branchiootorenal syndrome type 1 (EYA1)
437.	Branchiootorenal syndrome type 2 (SIX5)
438.	Breast cancer, male, susceptibility to (BRCA2)
439.	Breast cancer, RINT1 related (RINT1)
440.	Breast cancer, somatic (KRAS)
441.	Breast cancer, susceptibility to (PALB2)
442.	Breast cancer, susceptibility to (RECQL)
443.	Breast cancer, susceptibility to (BARD1)
444.	Breast cancer, susceptibility to (XRCC3)
445.	Breast-ovarian cancer (BRCA1)
446.	Breast-ovarian cancer (RAD51C)

447.	Breast-ovarian cancer, familial, susceptibility to, type 4 (RAD51D)
448.	Breast-ovarian cancer, familial, type 2 (BRCA2)
449.	Brittle cornea syndrome (ZNF469)
450.	Brody myopathy (ATP2A1)
451.	Bronchiectasis with or without elevated sweat chloride type 2 (SCNN1A)
452.	Brown-Vialetto-Van Laere syndrome 1 (SLC52A3)
453.	Brown-Vialetto-Van Laere syndrome type 2 (SLC52A2)
454.	Brugada syndrome type 1 (SCN5A)
455.	Brugada syndrome type 2 (GPD1L)
456.	Brugada syndrome type 3 (CACNA1C)
457.	Brugada syndrome type 4 (CACNB2)
458.	Brugada syndrome type 5 (SCN1B)
459.	Brugada syndrome type 6 (KCNE3)
460.	Brugada syndrome type 7 (SCN3B)
461.	Brugada syndrome type 8 (HCN4)
462.	Brugada syndrome type 9 (SLMAP)
463.	Brunner syndrome (MAOA)
464.	Budd-Chiari syndrome (F5)
465.	Buschke-Ollendorff syndrome (LEMD3)
466.	Butyrylcholinesterase deficiency (BCHE)
467.	C syndrome (CD96)
468.	C1q deficiency (C1QA)
469.	C2 deficiency (C2)
470.	C3 deficiency (C3)
471.	C5 deficiency (C5)
472.	C7 deficiency (C7)
473.	CADASIL (NOTCH3)
474.	CALR, selective sequencing of exon 9 (CALR)
475.	Campomelic dysplasia (SOX9)
476.	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome (PRG4)
477.	Camurati-Engelmann disease (TGFB1)
478.	Canavan disease (ASPA)

479.	Cantu syndrome (ABCC9)
480.	Capillary malformation-arteriovenous malformation (RASA1)
481.	CAPOS syndrome (ATP1A3)
482.	CARASIL (HTRA1)
483.	Carbamoylphosphate synthetase I deficiency (CPS1)
484.	Carcinoid tumor of lung, somatic (MEN1)
485.	Carcinoid tumors, intestinal (SDHD)
486.	Cardiac defects, CNOT3 related (CNOT3)
487.	Cardiac defects, PPP1R8 related (PPP1R8)
488.	Cardiac valvular dysplasia, X-linked (FLNA)
489.	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency (SCO2)
490.	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency type 2 (COX15)
491.	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency type 3 (COA5)
492.	Cardiofaciocutaneous syndrome (KRAS)
493.	Cardiofaciocutaneous syndrome (BRAF)
494.	Cardiofaciocutaneous syndrome type 3 (MAP2K1)
495.	Cardiofaciocutaneous syndrome type 4 (MAP2K2)
496.	Cardiomyopathy, apical hypertrophic, and neuropathy, MT-ATP8 related (NGS Sequencing: MT-ATP8)
497.	Cardiomyopathy, dilated (MYBPC3)
498.	Cardiomyopathy, dilated type 1 (CRYAB)
499.	Cardiomyopathy, dilated type 1A (LMNA)
500.	Cardiomyopathy, dilated type 1AA (ACTN2)
501.	Cardiomyopathy, dilated type 1BB (DSG2)
502.	Cardiomyopathy, dilated type 1C (LDB3)
503.	Cardiomyopathy, dilated type 1CC (NEXN)
504.	Cardiomyopathy, dilated type 1D (TNNT2)
505.	Cardiomyopathy, dilated type 1DD (RBM20)
506.	Cardiomyopathy, dilated type 1E (SCN5A)
507.	Cardiomyopathy, dilated type 1EE (MYH6)
508.	Cardiomyopathy, dilated type 1G (NGS Sequencing: TTN)
509.	Cardiomyopathy, dilated type 1GG (SDHA)
510.	Cardiomyopathy, dilated type 1HH (BAG3)

511.	Cardiomyopathy, dilated type 1I (DES)
512.	Cardiomyopathy, dilated type 1J (EYA4)
513.	Cardiomyopathy, dilated type 1KK (MYPN)
514.	Cardiomyopathy, dilated type 1L (SGCD)
515.	Cardiomyopathy, dilated type 1LL (PRDM16)
516.	Cardiomyopathy, dilated type 1M (CSRP3)
517.	Cardiomyopathy, dilated type 1N (TCAP)
518.	Cardiomyopathy, dilated type 1O (ABCC9)
519.	Cardiomyopathy, dilated type 1P (PLN)
520.	Cardiomyopathy, dilated type 1R (ACTC1)
521.	Cardiomyopathy, dilated type 1S (MYH7)
522.	Cardiomyopathy, dilated type 1T (TMPO)
523.	Cardiomyopathy, dilated type 1U (PSEN1)
524.	Cardiomyopathy, dilated type 1V (PSEN2)
525.	Cardiomyopathy, dilated type 1W (VCL)
526.	Cardiomyopathy, dilated type 1X (FKTN)
527.	Cardiomyopathy, dilated type 1Y (TPM1)
528.	Cardiomyopathy, dilated type 1Z (TNNC1)
529.	Cardiomyopathy, dilated type 2A (TNNI3)
530.	Cardiomyopathy, dilated type 2B (GATAD1)
531.	Cardiomyopathy, dilated type 3B (NGS Sequencing: DMD)
532.	Cardiomyopathy, dilated with ataxia (DNAJC19)
533.	Cardiomyopathy, dilated with hypergonadotropic hypogonadism (LMNA)
534.	Cardiomyopathy, dilated with woolly hair and keratoderma (DSP)
535.	Cardiomyopathy, familial hypertrophic (CAV3)
536.	Cardiomyopathy, familial hypertrophic type 1 (MYH7)
537.	Cardiomyopathy, familial hypertrophic type 10 (MYL2)
538.	Cardiomyopathy, familial hypertrophic type 11 (ACTC1)
539.	Cardiomyopathy, familial hypertrophic type 12 (CSRP3)
540.	Cardiomyopathy, familial hypertrophic type 16 (MYOZ2)
541.	Cardiomyopathy, familial hypertrophic type 17 (JPH2)
542.	Cardiomyopathy, familial hypertrophic type 19 (CALR3)

543.	Cardiomyopathy, familial hypertrophic type 2 (TNNT2)
544.	Cardiomyopathy, familial hypertrophic type 3 (TPM1)
545.	Cardiomyopathy, familial hypertrophic type 4 (MYBPC3)
546.	Cardiomyopathy, familial hypertrophic type 6 (PRKAG2)
547.	Cardiomyopathy, familial hypertrophic type 7 (TNNI3)
548.	Cardiomyopathy, familial hypertrophic type 8 (MYL3)
549.	Cardiomyopathy, familial hypertrophic type 9 (NGS Sequencing: TTN)
550.	Cardiomyopathy, familial restrictive type 1 (TNNI3)
551.	Cardiomyopathy, fatal, MT-TI related (NGS Sequencing: MT-TI)
552.	Cardiomyopathy, hypertrophic, midventricular, digenic (MYLK2)
553.	Cardiomyopathy, hypertrophic, MT-TG related (NGS Sequencing: MT-TG)
554.	Cardiomyopathy, hypertrophic, type 18 (PLN)
555.	Cardiomyopathy, hypertrophic, type 24 (LDB3)
556.	Cardiomyopathy, idiopathic dilated, mitochondrial, MT-TH related (NGS Sequencing: MT-TH)
557.	Cardiomyopathy, infantile hypertrophic, MT-ATP8 related (NGS Sequencing: MT-ATP8)
558.	Cardiomyopathy, left ventricular noncompaction, MYH7B related (MYH7B)
559.	Carney complex type 1 (PRKAR1A)
560.	Carnitine deficiency (SLC22A5)
561.	Carnitine palmitoyltransferase 1A deficiency (CPT1A)
562.	Carnitine palmitoyltransferase 1B deficiency (CPT1B)
563.	Carnitine palmitoyltransferase 2 deficiency, infantile (CPT2)
564.	Carnitine palmitoyltransferase 2 deficiency, lethal neonatal (CPT2)
565.	Carnitine-acylcarnitine translocase deficiency (SLC25A20)
566.	Carotid intimal medial thickness type 1 (PPARG)
567.	Carpenter syndrome (RAB23)
568.	Carpenter syndrome type 2 (MEGF8)
569.	Cartilage-hair hypoplasia (RMRP)
570.	Cataract 11, multiple types (PITX3)
571.	Cataract type 17, multiple types (CRYBB1)
572.	Cataract type 23 (CRYBA4)
573.	Cataract type 41 (WFS1)
574.	Cataract type 43 (UNC45B)

575.	Cataract, autosomal dominant (GCNT2)
576.	Cataract, autosomal recessive congenital nuclear type 2 (CRYBB3)
577.	Cataract, autosomal recessive congenital type 1 (CRYAA)
578.	Cataract, autosomal recessive congenital type 2 (FYCO1)
579.	Cataract, autosomal recessive congenital type 4 (TDRD7)
580.	Cataract, autosomal recessive type 38 (AGK)
581.	Cataract, congenital (SORD)
582.	Cataract, congenital, associated with Marinesco-Sjogren Syndrome (SIL1)
583.	Cataract, cortical pulverulent, late-onset (LIM2)
584.	Cataract, lamellar (HSF4)
585.	Cataract, posterior polar type 2 (CRYAB)
586.	Cataract, pulverulent or cerulean, with or without microcornea (MAF)
587.	Cataract, X-linked (NHS)
588.	Cataract-microcornea syndrome (GJA8)
589.	Cataracts with facial dysmorphism and neuropathy (CTDP1)
590.	Catechol-o-methyltransferase deficiency (COMT)
591.	Cell cycle disorder, CDC20 related (CDC20)
592.	Central core disease (NGS Sequencing: RYR1)
593.	Central hypoventilation syndrome with or without Hirschsprung disease (PHOX2B)
594.	Central hypoventilation syndrome, congenital (GDNF)
595.	Central hypoventilation syndrome, congenital (ASCL1)
596.	Central hypoventilation syndrome, congenital (RET)
597.	Central hypoventilation syndrome, congenital (ECE1)
598.	Central hypoventilation syndrome, congenital (MECP2)
599.	Central hypoventilation syndrome, congenital (ZEB2)
600.	Central hypoventilation syndrome, congenital (GFRA1)
601.	Central hypoventilation syndrome, congenital (PHOX2A)
602.	Central hypoventilation syndrome, congenital (EDN3)
603.	Central hypoventilation syndrome, congenital (BDNF)
604.	Centronuclear myopathy type 1 (MTMR14)
605.	Centronuclear myopathy type 3 (MYF6)
606.	Centronuclear myopathy type 4 (CCDC78)

607.	Cerebellar ataxia (CP)
608.	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion type 3 (CA8)
609.	Cerebellar ataxia with deafness and narcolepsy, autosomal recessive (DNMT1)
610.	Cerebellar ataxia with mental retardation and dysequilibrium syndrome type 2 (WDR81)
611.	Cerebellar ataxia with spasticity (GBA2)
612.	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome type 4 (ATP8A2)
613.	Cerebellar ataxia, nonprogressive, with mental retardation (CAMTA1)
614.	Cerebellar ataxia, SNX14 related (SNX14)
615.	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion type 1 (VLDLR)
616.	Cerebellar-retinal degeneration, infantile (ACO2)
617.	Cerebral amyloid angiopathy (CST3)
618.	Cerebral amyloid angiopathy, APP related (APP)
619.	Cerebral cavernous malformations type 1 (KRIT1)
620.	Cerebral cavernous malformations type 2 (CCM2)
621.	Cerebral cavernous malformations type 3 (PDCD10)
622.	Cerebral dysgenesis, neuropathy, ichthyosis, palmoplantar keratoderma syndrome (SNAP29)
623.	Cerebral palsy type 1, spastic quadriplegic (GAD1)
624.	Cerebral palsy type 2, spastic quadriplegic (KANK1)
625.	Cerebrooculofacioskeletal syndrome type 1 (ERCC6)
626.	Cerebrooculofacioskeletal syndrome type 4 (ERCC1)
627.	Cerebrotendinous xanthomatosis (CYP27A1)
628.	Ceroid lipofuscinosis neuronal type 1 (PPT1)
629.	Ceroid lipofuscinosis neuronal type 10 (CTSD)
630.	Ceroid lipofuscinosis neuronal type 11 (GRN)
631.	Ceroid lipofuscinosis neuronal type 2 (TPP1)
632.	Ceroid lipofuscinosis neuronal type 3 (CLN3)
633.	Ceroid lipofuscinosis neuronal type 4 (DNAJC5)
634.	Ceroid lipofuscinosis neuronal type 5 (CLN5)
635.	Ceroid lipofuscinosis neuronal type 6 (CLN6)
636.	Ceroid lipofuscinosis neuronal type 7 (MFSD8)
637.	Ceroid lipofuscinosis neuronal type 8 (CLN8)
638.	Cervical cancer, somatic (FGFR3)

639.	Cervical dystonia (CIZ1)
640.	Chanarin-Dorfman syndrome (ABHD5)
641.	Charcot-Marie-Tooth disease, axonal type 20 (NGS Sequencing: DYNC1H1)
642.	CHARGE syndrome (CHD7)
643.	Chediak-Higashi syndrome (NGS Sequencing: LYST)
644.	CHILD syndrome (NSDHL)
645.	CHIME syndrome (PIGL)
646.	Chloramphenicol resistance, MT-RNR2 related (NGS Sequencing: MT-RNR2)
647.	Choanal atresia and lymphedema (PTPN14)
648.	Cholestasis benign recurrent intrahepatic type 2 (ABCB11)
649.	Cholestasis intrahepatic, of pregnancy, type 3 (ABCB4)
650.	Cholestasis progressive intrahepatic type 1 (ATP8B1)
651.	Cholestasis progressive intrahepatic type 2 (ABCB11)
652.	Cholestasis progressive intrahepatic type 3 (ABCB4)
653.	Cholestasis, benign recurrent intrahepatic (ATP8B1)
654.	Cholestasis, infantile, NR1H4 related (NR1H4)
655.	Cholestasis, intrahepatic, of pregnancy, type 1 (ATP8B1)
656.	Cholesteryl ester storage disease (LIPA)
657.	Chondrocalcinosis type 2 (ANKH)
658.	Chondrodysplasia punctata, X-linked dominant (EBP)
659.	Chondrodysplasia punctata, X-linked recessive (ARSE)
660.	Chondrodysplasia with joint dislocations, GPAPP type (IMPAD1)
661.	Chondrodysplasia, Blomstrand type (PTH1R)
662.	Chondrosarcoma, familial (EXT1)
663.	Chorea, hereditary benign (NKX2-1)
664.	Choreoacanthocytosis (NGS Sequencing: VPS13A)
665.	Choroidal dystrophy, central areolar type 2 (PRPH2)
666.	Choroideremia (CHM)
667.	Chrondrodysplasia, acromesomelic, with genital anomalies (BMPR1B)
668.	Chudley-McCullough syndrome (GPSM2)
669.	Chylomicron retention disease (SAR1B)
670.	Ciliogenesis related disorder (PTPN23)

671.	Citrin deficiency (SLC25A13)
672.	Citrullinemia (ASS1)
673.	CK syndrome (NSDHL)
674.	Cleidocranial dysplasia (RUNX2)
675.	Club foot (PITX1)
676.	CMT1A (PMP22)
677.	CMT1B (MPZ)
678.	CMT1C (LITAF)
679.	CMT1D (EGR2)
680.	CMT1E (PMP22)
681.	CMT1F (NEFL)
682.	CMT2A1 (KIF1B)
683.	CMT2A2 (MFN2)
684.	CMT2B (RAB7A)
685.	CMT2B1 (LMNA)
686.	CMT2B2 (MED25)
687.	CMT2C (TRPV4)
688.	CMT2D (GARS)
689.	CMT2E (NEFL)
690.	CMT2F (HSPB1)
691.	CMT2I (MPZ)
692.	CMT2J (MPZ)
693.	CMT2K (GDAP1)
694.	CMT2L (HSPB8)
695.	CMT2N (AARS)
696.	CMT2P (LRSAM1)
697.	CMT4, CTDP1 related (CTDP1)
698.	CMT4A (GDAP1)
699.	CMT4B1 (MTMR2)
700.	CMT4B2 (SBF2)
701.	CMT4C (SH3TC2)
702.	CMT4D (NDRG1)

703.	CMT4E (MPZ)
704.	CMT4E (EGR2)
705.	CMT4F (PRX)
706.	CMT4H (FGD4)
707.	CMT4J (FIG4)
708.	CMTDIF (GNB4)
709.	CMTRIB (KARS)
710.	CMTRID (COX6A1)
711.	CMTX1 (GJB1)
712.	CMTX4 (AIFM1)
713.	CMTX5 (PRPS1)
714.	CoA-2 4-dienoyl reductase 1 deficiency (DECR1)
715.	CoA-3-hydroxyacyl dehydrogenase deficiency (HADH)
716.	CoA-3-methylcrontonyl carboxylase 1 deficiency (MCCC1)
717.	CoA-3-methylcrontonyl carboxylase 2 deficiency (MCCC2)
718.	COACH syndrome (TMEM67)
719.	COACH syndrome (RPGRIP1L)
720.	COACH syndrome (CC2D2A)
721.	Coarctation of the aorta (MCTP2)
722.	Coat plus syndrome (CTC1)
723.	Cockayne syndrome type A (ERCC8)
724.	Cockayne syndrome type B (ERCC6)
725.	CODAS syndrome (LONP1)
726.	Coenzyme Q10 deficiency type 1 (COQ2)
727.	Coenzyme Q10 deficiency type 2 (PDSS1)
728.	Coenzyme Q10 deficiency type 3 (PDSS2)
729.	Coenzyme Q10 deficiency type 5 (COQ9)
730.	Coffin-Lowry syndrome (RPS6KA3)
731.	Coffin-Siris syndrome, SMARCE1 related (SMARCE1)
732.	Cohen syndrome (NGS Sequencing: VPS13B)
733.	Colchicine resistance (ABCB1)
734.	Cold autoinflammatory syndrome type 2 (NLRP12)

735.	Cold-induced sweating syndrome (CRLF1)
736.	Cold-induced sweating syndrome type 2 (CLCF1)
737.	Cole disease (ENPP1)
738.	Coloboma of optic nerve (PAX6)
739.	Coloboma, ocular, autosomal dominant (PAX6)
740.	Colobomatous microphthalmia (TENM1)
741.	Colon cancer, PPARG related, somatic (PPARG)
742.	Colorectal cancer, hereditary (NRAS)
743.	Colorectal cancer, hereditary nonpolyposis type 1 (MSH2)
744.	Colorectal cancer, hereditary nonpolyposis type 2 (MLH1)
745.	Colorectal cancer, hereditary nonpolyposis type 4 (PMS2)
746.	Colorectal cancer, hereditary nonpolyposis type 5 (MSH6)
747.	Colorectal cancer, hereditary nonpolyposis type 6 (TGFBR2)
748.	Colorectal cancer, hereditary nonpolyposis type 7 (MLH3)
749.	Colorectal cancer, hereditary nonpolyposis type 8 (EPCAM)
750.	Colorectal cancer, hereditary, susceptibility to (CCND1)
751.	Colorectal Cancer, resistance to cetuximab, EGFR related, somatic (EGFR)
752.	Colorectal cancer, somatic (EP300)
753.	Colorectal cancer, somatic (DCC)
754.	Colorectal cancer, somatic (CTNNB1)
755.	Colorectal cancer, somatic (FGFR3)
756.	Colorectal cancer, somatic (FLCN)
757.	Colorectal cancer, somatic (NRAS)
758.	Colorectal cancer, somatic (BRAF)
759.	Colorectal cancer, somatic (APC)
760.	Combined cellular and humoral immune defects with granulomas (RAG2)
761.	Combined D-2- and L-2-hydroxyglutaric aciduria (SLC25A1)
762.	Combined immunodeficiency, B cell-negative, T cell-negative, NK cell positive (RAG2)
763.	Combined immunodeficiency, X-linked, moderate (IL2RG)
764.	Combined malonic and methylmalonic aciduria (ACSF3)
765.	Combined oxidative phosphorylation deficiency type 1 (GFM1)
766.	Combined oxidative phosphorylation deficiency type 10 (MTO1)

767.	Combined oxidative phosphorylation deficiency type 11 (RMND1)
768.	Combined oxidative phosphorylation deficiency type 12 (EARS2)
769.	Combined oxidative phosphorylation deficiency type 13 (PNPT1)
770.	Combined oxidative phosphorylation deficiency type 14 (FARS2)
771.	Combined oxidative phosphorylation deficiency type 15 (MTFMT)
772.	Combined oxidative phosphorylation deficiency type 16 (MRPL44)
773.	Combined oxidative phosphorylation deficiency type 17 (ELAC2)
774.	Combined oxidative phosphorylation deficiency type 18 (SFXN4)
775.	Combined oxidative phosphorylation deficiency type 19 (LYRM4)
776.	Combined oxidative phosphorylation deficiency type 2 (MRPS16)
777.	Combined oxidative phosphorylation deficiency type 20 (VARS2)
778.	Combined oxidative phosphorylation deficiency type 21 (TARS2)
779.	Combined oxidative phosphorylation deficiency type 22 (ATP5A1)
780.	Combined oxidative phosphorylation deficiency type 23 (GTPBP3)
781.	Combined oxidative phosphorylation deficiency type 24 (NARS2)
782.	Combined oxidative phosphorylation deficiency type 25 (MARS2)
783.	Combined oxidative phosphorylation deficiency type 26 (TRMT5)
784.	Combined oxidative phosphorylation deficiency type 3 (TSFM)
785.	Combined oxidative phosphorylation deficiency type 4 (TUFM)
786.	Combined oxidative phosphorylation deficiency type 5 (MRPS22)
787.	Combined oxidative phosphorylation deficiency type 6 (AIFM1)
788.	Combined oxidative phosphorylation deficiency type 7 (C12ORF65)
789.	Combined oxidative phosphorylation deficiency type 8 (AARS2)
790.	Combined oxidative phosphorylation deficiency type 9 (MRPL3)
791.	Compton-North congenital myopathy (CNTN1)
792.	Cone-rod dystrophy (UNC119)
793.	Cone-rod dystrophy (AIPL1)
794.	Cone-rod dystrophy type 11 (RAX2)
795.	Cone-rod dystrophy type 12 (PROM1)
796.	Cone-rod dystrophy type 13 (RPGRIP1)
797.	Cone-rod dystrophy type 14 (GUCA1A)
798.	Cone-rod dystrophy type 15 (CDHR1)

799.	Cone-rod dystrophy type 17 (CD3G)
800.	Cone-rod dystrophy type 2 (CRX)
801.	Cone-rod dystrophy type 20 (POC1B)
802.	Cone-rod dystrophy type 3 (ABCA4)
803.	Cone-rod dystrophy type 4 (PDE6C)
804.	Cone-rod dystrophy type 5 (PITPNM3)
805.	Cone-rod dystrophy type 7 (RIMS1)
806.	Cone-rod dystrophy type 9 (ADAM9)
807.	Cone-rod dystrophy, C21orf2 related (C21orf2)
808.	Cone-rod dystrophy, X-linked type 3 (CACNA1F)
809.	Congenital bilateral absence of vas deferens (NGS Sequencing: CFTR)
810.	Congenital disorder of glycosylation, type Ip (ALG11)
811.	Congenital disorder of glycosylation, type Iq (SRD5A3)
812.	Congenital disorder of glycosylation, type Iw (STT3A)
813.	Congenital heart defects multiple types (TAB2)
814.	Congenital heart disease and transposition of the great arteries (FOXH1)
815.	Congenital muscular dystrophy and hypoglycosylation of α-dystroglycan (B3GALNT2)
816.	Congenital short-bowel syndrome (CLMP)
817.	Contractural arachnodactyly, congenital (NGS Sequencing: FBN2)
818.	Convulsions, benign familial infantile, 3 (SCN2A)
819.	Convulsions, familial infantile, with paroxysmal choreoathetosis (PRRT2)
820.	Coproporphyria (CPOX)
821.	Corneal dystrophy, epithelial basement membrane (TGFBI)
822.	Corneal dystrophy, posterior polymorphous, type 1 (VSX1)
823.	Corneal endothelial dystrophy type 2 (SLC4A11)
824.	Corneal intraepithelial dyskeratosis and ectodermal dysplasia (NLRP1)
825.	Corneal opacification and other ocular anomalies (PXDN)
826.	Cornelia de Lange syndrome type 1 (NIPBL)
827.	Cornelia de Lange syndrome type 2 (SMC1A)
828.	Cornelia de Lange syndrome type 3 (SMC3)
829.	Cornelia de Lange syndrome type 4 (RAD21)
830.	Cornelia de Lange syndrome type 5 (HDAC8)

831.	Coronary artery disease in familial hypercholesterolemia, protection against (ABCA1)
832.	Coronary heart disease, susceptibility to, type 6 (MMP3)
833.	Corpus callosum, agenesis of, with abnormal genitalia (ARX)
834.	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia (IGBP1)
835.	Cortical dysplasia, complex, with other brain malformations, type 1 (TUBB3)
836.	Cortical dysplasia, complex, with other brain malformations, type 5 (TUBB2A)
837.	Cortical dysplasia-focal epilepsy syndrome (CNTNAP2)
838.	Cortical malformations, occipital (LAMC3)
839.	Corticobasal Degeneration, CFL1 related (CFL1)
840.	Costello syndrome (HRAS)
841.	Coumarin resistance (VKORC1)
842.	Coumarin/Warfarin resistance due to CYP2C9 variants (CYP2C9)
843.	Cousin syndrome (TBX15)
844.	Cowden syndrome type 1 (PTEN)
845.	Cowden syndrome type 3 (SDHD)
846.	Cowden syndrome type 5 (PIK3CA)
847.	Cowden syndrome type 6 (AKT1)
848.	Craniodiaphyseal dysplasia, autosomal dominant (SOST)
849.	Cranioectodermal dysplasia type 1 (IFT122)
850.	Cranioectodermal dysplasia type 2 (WDR35)
851.	Cranioectodermal dysplasia type 3 (IFT43)
852.	Cranioectodermal dysplasia type 4 (WDR19)
853.	Craniofacial and neuro-developmental abnormalities (DISP1)
854.	Craniofacial and neuro-developmental abnormalities, JAG2 related (JAG2)
855.	Craniofacial anomalies and anterior segment dysgenesis syndrome (VSX1)
856.	Craniofacial-skeletal-dermatologic dysplasia (FGFR2)
857.	Craniofrontonasal syndrome (EFNB1)
858.	Craniometaphyseal dysplasia (ANKH)
859.	Craniosynostosis and dental anomalies (IL11RA)
860.	Craniosynostosis type 1 (TWIST1)
861.	Craniosynostosis type 2 (MSX2)
862.	Craniosynostosis type 3 (TCF12)

863.	Craniosynostosis type 4 (ERF)
864.	Craniosynostosis type 6 (ZIC1)
865.	Craniosynostosis, FGFR1 related (FGFR1)
866.	Craniosynostosis, nonspecific (FGFR2)
867.	Creatine deficiency syndrome X-linked (SLC6A8)
868.	Creatine phosphokinase, elevated serum (CAV3)
869.	Creutzfeldt-Jakob disease (PRNP)
870.	Crigler-Najjar syndrome, type 1 (UGT1A1)
871.	Crigler-Najjar syndrome, type 2 (UGT1A1)
872.	Crouzon syndrome (FGFR2)
873.	Crouzon syndrome with acanthosis nigricans (FGFR3)
874.	Cryptorchidism (RXFP2)
875.	Currarino syndrome (MNX1)
876.	Cutaneous telangiectasia and cancer syndrome, familial (NGS Sequencing: ATR)
877.	Cutis laxa type 1A, autosomal recessive (FBLN5)
878.	Cutis laxa type 1B, autosomal recessive (EFEMP2)
879.	Cutis laxa type 1C, autosomal recessive (LTBP4)
880.	Cutis laxa type 2, autosomal dominant (FBLN5)
881.	Cutis laxa type 2A, autosomal recessive (ATP6V0A2)
882.	Cutis laxa type 2B, autosomal recessive (PYCR1)
883.	Cutis laxa type 3A, autosomal recessive (ALDH18A1)
884.	Cutis laxa type 3B, autosomal recessive (PYCR1)
885.	Cutis laxa, autosomal dominant (ELN)
886.	Cyanosis, transient neonatal (HBG2)
887.	Cylindromatosis, familial (CYLD)
888.	CYP2C19 related poor drug metabolism (CYP2C19)
889.	Cystathioninuria (CTH)
890.	Cystic fibrosis (NGS Sequencing: CFTR)
891.	Cystic fibrosis, SLC6A14 related (SLC6A14)
892.	Cystinosis, nephropathic (CTNS)
893.	Cystinuria (SLC7A9)
894.	Cystinuria (SLC3A1)

895.	Cystinuria (PREPL)
896.	Cytochrome c oxidase 1 deficiency (NGS Sequencing: MT-CO1)
897.	Cytochrome c oxidase 2 deficiency (NGS Sequencing: MT-CO2)
898.	Cytochrome c oxidase 3 deficiency (NGS Sequencing: MT-CO3)
899.	Cytochrome P450 deficiency (CYP1A2)
900.	Czech dysplasia (COL2A1)
901.	D-2-hydroxyglutaric aciduria type 1 (D2HGDH)
902.	D-2-hydroxyglutaric aciduria type 2 (IDH2)
903.	D-bifunctional protein deficiency (HSD17B4)
904.	D-glyceric aciduria (GLYCTK)
905.	Dandy-Walker malformation and occipital cephaloceles, LAMC1 related (LAMC1)
906.	Danon disease (LAMP2)
907.	De Sanctis-Cacchione syndrome (ERCC6)
908.	Deafness and male infertility (STRC)
909.	Deafness and male infertility, CATSPER2 related (CATSPER2)
910.	Deafness with keratopachydermia and constrictions of fingers and toes (GJB2)
911.	Deafness, autosomal dominant type 1 (DIAPH1)
912.	Deafness, autosomal dominant type 10 (EYA4)
913.	Deafness, autosomal dominant type 11 (MYO7A)
914.	Deafness, autosomal dominant type 12 (TECTA)
915.	Deafness, autosomal dominant type 13 (COL11A2)
916.	Deafness, autosomal dominant type 15 (POU4F3)
917.	Deafness, autosomal dominant type 17 (MYH9)
918.	Deafness, autosomal dominant type 20 (ACTG1)
919.	Deafness, autosomal dominant type 22 (MYO6)
920.	Deafness, autosomal dominant type 23 (SIX1)
921.	Deafness, autosomal dominant type 25 (SLC17A8)
922.	Deafness, autosomal dominant type 28 (GRHL2)
923.	Deafness, autosomal dominant type 2A (KCNQ4)
924.	Deafness, autosomal dominant type 2B (GJB3)
925.	Deafness, autosomal dominant type 36 (TMC1)
926.	Deafness, autosomal dominant type 39, with dentinogenesis type 1 (DSPP)

927.	Deafness, autosomal dominant type 3A (GJB2)
928.	Deafness, autosomal dominant type 3B (GJB6)
929.	Deafness, autosomal dominant type 4 (MYH14)
930.	Deafness, autosomal dominant type 40 (CRYM)
931.	Deafness, autosomal dominant type 44 (CCDC50)
932.	Deafness, autosomal dominant type 48 (MYO1A)
933.	Deafness, autosomal dominant type 4B (CEACAM16)
934.	Deafness, autosomal dominant type 5 (DFNA5)
935.	Deafness, autosomal dominant type 50 (MIR96)
936.	Deafness, autosomal dominant type 52 (POU4F3)
937.	Deafness, autosomal dominant type 6 (WFS1)
938.	Deafness, autosomal dominant type 64 (DIABLO)
939.	Deafness, autosomal dominant type 65 (TBC1D24)
940.	Deafness, autosomal dominant type 9 (COCH)
941.	Deafness, autosomal recessive (GJB3)
942.	Deafness, autosomal recessive (SUN1)
943.	Deafness, autosomal recessive type 15 (GIPC3)
944.	Deafness, autosomal recessive type 16 (STRC)
945.	Deafness, autosomal recessive type 18 (USH1C)
946.	Deafness, autosomal recessive type 1A (GJB2)
947.	Deafness, autosomal recessive type 1B (GJB6)
948.	Deafness, autosomal recessive type 2 (MYO7A)
949.	Deafness, autosomal recessive type 22 (OTOA)
950.	Deafness, autosomal recessive type 23 (PCDH15)
951.	Deafness, autosomal recessive type 24 (RDX)
952.	Deafness, autosomal recessive type 25 (GRXCR1)
953.	Deafness, autosomal recessive type 28 (TRIOBP)
954.	Deafness, autosomal recessive type 29 (CLDN14)
955.	Deafness, autosomal recessive type 3 (MYO15A)
956.	Deafness, autosomal recessive type 30 (MYO3A)
957.	Deafness, autosomal recessive type 31 (DFNB31)
958.	Deafness, autosomal recessive type 35 (ESRRB)

959.	Deafness, autosomal recessive type 36 (ESPN)
960.	Deafness, autosomal recessive type 39 (HGF)
961.	Deafness, autosomal recessive type 4 (FOXI1)
962.	Deafness, autosomal recessive type 42 (ILDR1)
963.	Deafness, autosomal recessive type 48 (CIB2)
964.	Deafness, autosomal recessive type 49 (MARVELD2)
965.	Deafness, autosomal recessive type 53 (COL11A2)
966.	Deafness, autosomal recessive type 59 (DFNB59)
967.	Deafness, autosomal recessive type 6 (TMIE)
968.	Deafness, autosomal recessive type 61 (SLC26A5)
969.	Deafness, autosomal recessive type 63 (LRTOMT)
970.	Deafness, autosomal recessive type 66 (DCDC2)
971.	Deafness, autosomal recessive type 67 (LHFPL5)
972.	Deafness, autosomal recessive type 7 (TMC1)
973.	Deafness, autosomal recessive type 70 (PNPT1)
974.	Deafness, autosomal recessive type 74 (MSRB3)
975.	Deafness, autosomal recessive type 76 (SYNE4)
976.	Deafness, autosomal recessive type 77 (LOXHD1)
977.	Deafness, autosomal recessive type 79 (TPRN)
978.	Deafness, autosomal recessive type 8/10 (TMPRSS3)
979.	Deafness, autosomal recessive type 84 (PTPRQ)
980.	Deafness, autosomal recessive type 86 (TBC1D24)
981.	Deafness, autosomal recessive type 89 (KARS)
982.	Deafness, autosomal recessive type 9 (OTOF)
983.	Deafness, autosomal recessive type 91 (SERPINB6)
984.	Deafness, autosomal recessive type 93 (CABP2)
985.	Deafness, autosomal recessive, type 12 (NGS Sequencing: CDH23)
986.	Deafness, congenital with inner ear agenesis, microtia, and microdontia (FGF3)
987.	Deafness, dystonia, and cerebral hypomyelination, X-linked (BCAP31)
988.	Deafness, nonsyndromic, sensorineural, mitochondrial (NGS Sequencing: MT-RNR1)
989.	Deafness, X-linked type 1 (PRPS1)
990.	Deafness, X-linked type 2 (POU3F4)

991.	Deafness, X-linked type 4 (SMPX)
992.	Deafness, X-linked type 5 (AIFM1)
993.	Deafness, X-linked type 6 (COL4A6)
994.	Dehydrated hereditary stomatocytosis (PIEZO1)
995.	Dejerine-Sottas disease (EGR2)
996.	Dejerine-Sottas disease (PRX)
997.	Dejerine-Sottas disease (PMP22)
998.	Dejerine-Sottas disease (MPZ)
999.	Dejerine-Sottas disease (GJB1)
1000.	Delta-beta thalassemia (HBB)
1001.	Dementia, familial, British type (ITM2B)
1002.	Dementia, familial, Danish type (ITM2B)
1003.	Dementia, frontotemporal (GRN)
1004.	Dementia, frontotemporal (MAPT)
1005.	Dementia, frontotemporal (TARDBP)
1006.	Dementia, frontotemporal (PSEN1)
1007.	Dementia, Lewy body (SNCA)
1008.	Dent disease (CLCN5)
1009.	Dent disease type 2 (OCRL)
1010.	Dentatorubral-pallidoluysian atrophy (ATN1)
1011.	Dentin dysplasia, type 2 (DSPP)
1012.	Dentinogenesis imperfecta, Shields type 2 (DSPP)
1013.	Dentinogenesis imperfecta, Shields type 3 (DSPP)
1014.	Dermatitis, atopic type 2 (FLG)
1015.	Dermatopathia pigmentosa reticularis (KRT14)
1016.	Desbuquois dysplasia type 1 (CANT1)
1017.	Desbuquois dysplasia type 2 (XYLT1)
1018.	Desmoid disease, hereditary (APC)
1019.	Desmosterolosis (DHCR24)
1020.	Developmental delay and microcephaly, SLC1A4 related (SLC1A4)
1021.	Developmental delay, GNAQ related (GNAQ)
1022.	Developmental delay, KMT2C related (NGS Sequencing: KMT2C)

1023.	DI-CMTB (DNM2)
1024.	DI-CMTC (YARS)
1025.	DI-CMTD (MPZ)
1026.	Diabetes insipidus, nephrogenic, autosomal (AQP2)
1027.	Diabetes insipidus, nephrogenic, X-linked (AVPR2)
1028.	Diabetes insipidus, neurohypophyseal (AVP)
1029.	Diabetes mellitus type 1 (INS)
1030.	Diabetes mellitus, insulin-dependent type 20 (HNF1A)
1031.	Diabetes mellitus, insulin-resistant with acanthosis nigricans (INSR)
1032.	Diabetes mellitus, neonatal (GLIS3)
1033.	Diabetes mellitus, noninsulin-dependent (KCNJ11)
1034.	Diabetes mellitus, noninsulin-dependent (AKT2)
1035.	Diabetes mellitus, noninsulin-dependent (ABCC8)
1036.	Diabetes mellitus, permanent neonatal (ABCC8)
1037.	Diabetes mellitus, transient neonatal type 2 (ABCC8)
1038.	Diabetes, IGF2 related (IGF2)
1039.	Diamond Blackfan anemia type 15 with mandibulofacial dysostosis (RPS28)
1040.	Diamond-Blackfan anemia type 1 (RPS19)
1041.	Diamond-Blackfan anemia type 10 (RPS26)
1042.	Diamond-Blackfan anemia type 11 (RPL26)
1043.	Diamond-Blackfan anemia type 12 (RPL15)
1044.	Diamond-Blackfan anemia type 13 (RPS29)
1045.	Diamond-Blackfan anemia type 14 with mandibulofacial dysostosis (TSR2)
1046.	Diamond-blackfan anemia type 3 (RPS24)
1047.	Diamond-Blackfan anemia type 4 (RPS17)
1048.	Diamond-Blackfan anemia type 5 (RPL35A)
1049.	Diamond-Blackfan anemia type 6 (RPL5)
1050.	Diamond-Blackfan anemia type 7 (RPL11)
1051.	Diamond-Blackfan anemia type 8 (RPS7)
1052.	Diamond-Blackfan anemia type 9 (RPS10)
1053.	Diaphragmatic hernia type 3 (ZFPM2)
1054.	Diaphyseal medullary stenosis with malignant fibrous histiocytoma (MTAP)

1055.	Diarrhea type 1, secretory chloride, congenital (SLC26A3)
1056.	Diarrhea type 2 with microvillus atrophy (MYO5B)
1057.	Diarrhea type 4, malabsorptive, congenital (NEUROG3)
1058.	Diarrhea type 6 (GUCY2C)
1059.	DiGeorge syndrome (TBX1)
1060.	Dihydropyrimidine dehydrogenase deficiency (DPYD)
1061.	Dihydropyrimidinuria (DPYS)
1062.	Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis (DSP)
1063.	Dimethylglycine dehydrogenase deficiency (DMGDH)
1064.	Disorders of sex development with cleft palate (FOXF2)
1065.	Donnai-Barrow syndrome (NGS Sequencing: LRP2)
1066.	DOOR syndrome (TBC1D24)
1067.	Dopamine beta-hydroxylase (DBH) deficiency (DBH)
1068.	Doyne honeycob retinal dystrophy (EFEMP1)
1069.	Dravet syndrome (GABRG2)
1070.	Dravet syndrome (SCN2A)
1071.	Dravet syndrome, modifier of (SCN9A)
1072.	Duane Retraction syndrome (SALL4)
1073.	Duane Retraction syndrome (CHN1)
1074.	Dubin-Johnson syndrome (ABCC2)
1075.	Dyggve-Melchior-Clausen disease (DYM)
1076.	Dysautonomia, FRRS1L-related (FRRS1L)
1077.	Dyschromatosis symmetrica hereditaria (ADAR)
1078.	Dyschromatosis universalis hereditaria type 3 (ABCB6)
1079.	Dyserythropoietic anemia (COX4I2)
1080.	Dyserythropoietic anemia, congenital, type 1B (C15orf41)
1081.	Dyserythropoietic anemia, congenital, type 3 (KIF23)
1082.	Dyserythropoietic anemia, congenital, type 4 (KLF1)
1083.	Dyskeratosis congenita, autosomal dominant type 1 (TERC)
1084.	Dyskeratosis congenita, autosomal recessive type 1 (NOP10)
1085.	Dyskeratosis congenita, autosomal recessive type 2 (NHP2)
1086.	Dyskeratosis congenita, autosomal recessive type 4/ autosomal dominant type 2 (TERT)

1087.	Dyskeratosis congenita, autosomal recessive type 5 (RTEL1)
1088.	Dyskeratosis congenita, autosomal recessive type 6 (PARN)
1089.	Dyskeratosis congenita, autosomal recessive type 7 (ACD)
1090.	Dyskeratosis congenita, X-linked (DKC1)
1091.	Dyskinesia, familial, with facial myokymia (ADCY5)
1092.	Dyslexia (PCDH11X)
1093.	Dysmorphism, HMG20B related (HMG20B)
1094.	Dysprothrombinemia (F2)
1095.	Dyssegmental dysplasia, Silverman-Handmaker type (NGS Sequencing: HSPG2)
1096.	Dystonia juvenile-onset (ACTB)
1097.	Dystonia, DOPA-responsive, autosomanl recessive (SPR)
1098.	Dystonia-deafness syndrome (TIMM8A)
1099.	DYT1 (TOR1A)
1100.	DYT10 (PRRT2)
1101.	DYT11 (SGCE)
1102.	DYT11, DRD2 related (DRD2)
1103.	DYT12 (ATP1A3)
1104.	DYT16 (PRKRA)
1105.	DYT18 (SLC2A1)
1106.	DYT2 (HPCA)
1107.	DYT23 (CACNA1B)
1108.	DYT24 (ANO3)
1109.	DYT25 (GNAL)
1110.	DYT26, myoclonic (KCTD17)
1111.	DYT27 (COL6A3)
1112.	DYT3 (TAF1)
1113.	DYT4 (TUBB4A)
1114.	DYT5A (GCH1)
1115.	DYT6 (THAP1)
1116.	DYT8 (SLC2A1)
1117.	Early infantile epileptic encephalopathy type 1 (ARX)
1118.	Early infantile epileptic encephalopathy type 10 (PNKP)

1119.	Early infantile epileptic encephalopathy type 11 (SCN2A)
1120.	Early infantile epileptic encephalopathy type 12 (PLCB1)
1121.	Early infantile epileptic encephalopathy type 13 (SCN8A)
1122.	Early infantile epileptic encephalopathy type 14 (KCNT1)
1123.	Early infantile epileptic encephalopathy type 15 (ST3GAL3)
1124.	Early infantile epileptic encephalopathy type 16 (TBC1D24)
1125.	Early infantile epileptic encephalopathy type 17 (GNAO1)
1126.	Early infantile epileptic encephalopathy type 19 (GABRA1)
1127.	Early infantile epileptic encephalopathy type 2 (CDKL5)
1128.	Early infantile epileptic encephalopathy type 20 (PIGA)
1129.	Early infantile epileptic encephalopathy type 21 (NECAP1)
1130.	Early infantile epileptic encephalopathy type 23 (DOCK7)
1131.	Early infantile epileptic encephalopathy type 24 (HCN1)
1132.	Early infantile epileptic encephalopathy type 25 (SLC13A5)
1133.	Early infantile epileptic encephalopathy type 26 (KCNB1)
1134.	Early infantile epileptic encephalopathy type 27 (GRIN2B)
1135.	Early infantile epileptic encephalopathy type 28 (WWOX)
1136.	Early infantile epileptic encephalopathy type 29 (AARS)
1137.	Early infantile epileptic encephalopathy type 3 (SLC25A22)
1138.	Early infantile epileptic encephalopathy type 30 (SIK1)
1139.	Early infantile epileptic encephalopathy type 31 (DNM1)
1140.	Early infantile epileptic encephalopathy type 32 (KCNA2)
1141.	Early infantile epileptic encephalopathy type 33 (EEF1A2)
1142.	Early infantile epileptic encephalopathy type 4 (STXBP1)
1143.	Early infantile epileptic encephalopathy type 40 (GUF1)
1144.	Early infantile epileptic encephalopathy type 45 (GABRB1)
1145.	Early infantile epileptic encephalopathy type 47 (FGF12)
1146.	Early infantile epileptic encephalopathy type 5 (SPTAN1)
1147.	Early infantile epileptic encephalopathy type 6 (SCN1A)
1148.	Early infantile epileptic encephalopathy type 7 (KCNQ2)
1149.	Early infantile epileptic encephalopathy type 8 (ARHGEF9)
1150.	Early infantile epileptic encephalopathy type 9 (PCDH19)

1151.	Early onset glaucoma, phenotype modifier of, COL15A1 related (COL15A1)
1152.	Ectodactyly, ectodermal dysplasia, and cleft lip/palate syndrome type 3 (TP63)
1153.	Ectodermal dysplasia type 4, hair/nail type (KRT85)
1154.	Ectodermal dysplasia, ectrodactyly, and macular dystrophy (CDH3)
1155.	Ectodermal dysplasia, hidrotic (GJB6)
1156.	Ectodermal dysplasia, hypohidrotic, autosomal recessive (EDAR)
1157.	Ectodermal dysplasia, hypohidrotic, autosomal recessive (EDARADD)
1158.	Ectodermal dysplasia, hypohidrotic, with immune deficiency (IKBKG)
1159.	Ectodermal dysplasia, hypohidrotic, X-linked (EDA)
1160.	Ectodermal dysplasia/skin fragility syndrome (PKP1)
1161.	Ectopia lentis et pupillae (ADAMTSL4)
1162.	Ectopia lentis, familial (NGS Sequencing: FBN1)
1163.	Ectopia lentis, isolated, autosomal recessive (ADAMTSL4)
1164.	Efavirenz, poor metabolism of (CYP2B6)
1165.	EGFR somatic Hotspot: c.2573T>G, p.L858R (EGFR)
1166.	EGFR, selective sequencing of exons 18-21 (EGFR)
1167.	Ehlers-Danlos syndrome type 1/2 (COL5A2)
1168.	Ehlers-Danlos syndrome type 1/2 (COL5A1)
1169.	Ehlers-Danlos syndrome type 3 (COL3A1)
1170.	Ehlers-Danlos syndrome type 3 (TNXB)
1171.	Ehlers-Danlos syndrome type 4 (COL5A1)
1172.	Ehlers-Danlos syndrome type 4 (COL3A1)
1173.	Ehlers-Danlos syndrome type 6 (PLOD1)
1174.	Ehlers-Danlos syndrome type 7A (COL1A1)
1175.	Ehlers-Danlos syndrome type 7B (COL1A2)
1176.	Ehlers-Danlos syndrome type 7C (ADAMTS2)
1177.	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss (FKBP14)
1178.	Ehlers-Danlos syndrome, musculocontractural type 1 (CHST14)
1179.	Ehlers-Danlos syndrome, musculocontractural type 2 (DSE)
1180.	Ehlers-Danlos syndrome, progeroid type 1 (B4GALT7)
1181.	Ehlers-Danlos syndrome, progeroid type, type 2 (B3GALT6)
1182.	Ellis-van Creveld syndrome (EVC)

1183.	Ellis-van Creveld syndrome (EVC2)
1184.	Emberger syndrome (GATA2)
1185.	Emery-Dreifuss muscular dystrophy type 1 (EMD)
1186.	Emery-Dreifuss muscular dystrophy type 2 (LMNA)
1187.	Emery-Dreifuss muscular dystrophy type 4 (NGS Sequencing: SYNE1)
1188.	Emery-Dreifuss muscular dystrophy type 5 (NGS Sequencing: SYNE2)
1189.	Emery-Dreifuss muscular dystrophy type 6 (FHL1)
1190.	Encephalomyopathy, mitochondrial, MT-TL2 related (NGS Sequencing: MT-TL2)
1191.	Encephalomyopathy, mitochondrial, MT-TR related (NGS Sequencing: MT-TR)
1192.	Encephalopathy acute necrotizing type 1 (RANBP2)
1193.	Encephalopathy lethal, due to defective mitochondrial peroxisomal fission (DNM1L)
1194.	Encephalopathy mitochondrial (VDAC1)
1195.	Encephalopathy mitochondrial with proximal renal tubulopathy due to cytochrome c oxidase deficiency (COX10)
1196.	Encephalopathy neonatal severe (MECP2)
1197.	Encephalopathy thiamine-responsive (SLC19A3)
1198.	Encephalopathy, familial, with neuroserpin inclusion bodies (SERPINI1)
1199.	Encephalopathy, mitochondrial, MT-TW related (NGS Sequencing: MT-TW)
1200.	Encephalopathy, progressive, with or without lipodystrophy (BSCL2)
1201.	Endometrial cancer, familial, MSH6 related (MSH6)
1202.	Endometrial carcinoma, somatic (CDH1)
1203.	Endometrioid carcinoma, ARID1A related, somatic (ARID1A)
1204.	Endplate acetylcholinesterase deficiency (COLQ)
1205.	Enterokinase deficiency (TMPRSS15)
1206.	Epidermal nevus, somatic (NRAS)
1207.	Epidermolysis bullosa dystrophica (COL7A1)
1208.	Epidermolysis bullosa dystrophica, autosomal recessive, modifier of (MMP1)
1209.	Epidermolysis bullosa junctionalis with pyloric atresia (ITGA6)
1210.	Epidermolysis bullosa junctionalis with pyloric atresia (ITGB4)
1211.	Epidermolysis bullosa simplex (KRT5)
1212.	Epidermolysis bullosa simplex with muscular dystrophy (NGS Sequencing: PLEC)
1213.	Epidermolysis bullosa simplex with pyloric atresia (NGS Sequencing: PLEC)
1214.	Epidermolysis bullosa simplex, autosomal recessive type 1 (KRT14)

1215.	Epidermolysis bullosa simplex, autosomal recessive type 2 (NGS Sequencing: DST)
1216.	Epidermolysis bullosa simplex, Dowling-Meara type (KRT14)
1217.	Epidermolysis bullosa simplex, Koebner type (KRT14)
1218.	Epidermolysis bullosa simplex, Ogna type (NGS Sequencing: PLEC)
1219.	Epidermolysis bullosa simplex, Weber-Cockayne type (KRT14)
1220.	Epidermolysis bullosa, generalized atrophic benign (NGS Sequencing: LAMA3)
1221.	Epidermolysis bullosa, junctional (LAMC2)
1222.	Epidermolysis bullosa, junctional (COL17A1)
1223.	Epidermolysis bullosa, junctional, Herlitz type (NGS Sequencing: LAMA3)
1224.	Epidermolysis bullosa, junctional, Herlitz type (LAMB3)
1225.	Epidermolysis bullosa, junctional, non-Herlitz type (LAMB3)
1226.	Epidermolysis bullosa, lethal acantholytic (DSP)
1227.	Epidermolysis bullosa, nonspecific, autosomal recessive (EXPH5)
1228.	Epidermolytic hyperkeratosis (KRT1)
1229.	Epidermolytic hyperkeratosis (KRT10)
1230.	Epidermolytic palmoplantar keratoderma (KRT9)
1231.	Epilepsy with neurodevelopmental defects (GRIN2A)
1232.	Epilepsy, childhood absence type 2 (GABRG2)
1233.	Epilepsy, childhood absence type 4, susceptibility to (GABRA1)
1234.	Epilepsy, childhood absence type 5 (GABRB3)
1235.	Epilepsy, childhood absence type 6, susceptibility to (CACNA1H)
1236.	Epilepsy, childhood absence, JRK related (JRK)
1237.	Epilepsy, familial focal with variable foci (DEPDC5)
1238.	Epilepsy, familial temporal lobe type 1 (LGI1)
1239.	Epilepsy, familial temporal lobe type 5 (CPA6)
1240.	Epilepsy, familial temporal lobe type 7 (NGS Sequencing: RELN)
1241.	Epilepsy, focal, SCN3A related (SCN3A)
1242.	Epilepsy, HCN2 related (HCN2)
1243.	Epilepsy, hearing loss, and mental retardation syndrome (SPATA5)
1244.	Epilepsy, idiopathic generalized type 10 (GABRD)
1245.	Epilepsy, idiopathic generalized type 11 (CLCN2)
1246.	Epilepsy, idiopathic generalized type 12 (SLC2A1)

1247.	Epilepsy, juvenile absence type 1 (EFHC1)
1248.	Epilepsy, nocturnal frontal lobe (KCNT1)
1249.	Epilepsy, nocturnal frontal lobe type 1 (CHRNA4)
1250.	Epilepsy, nocturnal frontal lobe type 3 (CHRNB2)
1251.	Epilepsy, nocturnal frontal lobe type 4 (CHRNA2)
1252.	Epilepsy, progressive myoclonic 4, with or without renal failure (SCARB2)
1253.	Epilepsy, progressive myoclonic type 5 (PRICKLE2)
1254.	Epilepsy, X-linked, with learning disabilities and behavior disorders (SYN1)
1255.	Epileptic encephalopathy, childhood-onset (CHD2)
1256.	Epileptic encephalopathy, Lennox-Gastaut type (MAPK10)
1257.	Epiphyseal dysplasia, multiple, type 1 (COMP)
1258.	Epiphyseal dysplasia, multiple, type 3 (COL9A3)
1259.	Epiphyseal dysplasia, multiple, type 5 (MATN3)
1260.	Epiphyseal dysplasia, multiple, with myopia and deafness (COL2A1)
1261.	Episodic ataxia type 1 (KCNA1)
1262.	Episodic ataxia type 2 (CACNA1A)
1263.	Episodic ataxia type 5 (CACNB4)
1264.	Episodic ataxia type 6 (SLC1A3)
1265.	Episodic pain syndrome type 2, familial (SCN10A)
1266.	Episodic pain syndrome type 3, familial (SCN11A)
1267.	Epstein syndrome (MYH9)
1268.	Erythermalgia, primary (SCN9A)
1269.	Erythrocyte lactate transporter defect (SLC16A1)
1270.	Erythrocytosis, familial type 1 (EPOR)
1271.	Erythrocytosis, familial type 3 (EGLN1)
1272.	Erythrocytosis, familial type 4 (EPAS1)
1273.	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis and hyper IgE (DSG1)
1274.	Erythrokeratodermia variabilis et progressive (GJB4)
1275.	Erythrokeratodermia variabilis et progressive (GJB3)
1276.	Esophageal cancer, somatic (TGFBR2)
1277.	Esophageal carcinoma, somatic (DCC)
1278.	Estrogen resistance (ESR1)

1279.	Ethylmalonic encephalopathy (ETHE1)
1280.	Exfoliation syndrome, susceptibility to (LOXL1)
1281.	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis (COX4I2)
1282.	Exostoses, multiple, type 1 (EXT1)
1283.	Exostoses, multiple, type 2 (EXT2)
1284.	Exudative vitreoretinopathy (FZD4)
1285.	Exudative vitreoretinopathy type 2 (NDP)
1286.	Exudative vitreoretinopathy type 5 (TSPAN12)
1287.	Fabry disease (GLA)
1288.	Facial paresis type 3 (HOXB1)
1289.	Faciogenital dysplasia (FGD1)
1290.	Facioscapulohumeral dystrophy-like phenotype, FAT1 related (FAT1)
1291.	Factor II deficiency (F2)
1292.	Factor V deficiency (F5)
1293.	Factor VII deficiency (F7)
1294.	Factor X deficiency (F10)
1295.	Factor XI deficiency (F11)
1296.	Factor XII deficiency (F12)
1297.	Factor XIIIA deficiency (F13A1)
1298.	Factor XIIIB deficiency (F13B)
1299.	Familial adenomatous polyposis coli (APC)
1300.	Familial adenomatous polyposis type 2 (MUTYH)
1301.	Familial adenomatous polyposis type 3 (NTHL1)
1302.	Familial adenomatous polyposis type 4 (MSH3)
1303.	Familial atrial fibrillation type 13 (SCN1B)
1304.	Familial hemiplegic migraine type 1 (CACNA1A)
1305.	Familial hemiplegic migraine type 2 (ATP1A2)
1306.	Familial hemiplegic migraine type 3 (SCN1A)
1307.	Familial infantile myoclonic epilepsy (TBC1D24)
1308.	Fanconi anemia type A (FANCA)
1309.	Fanconi anemia type B (FANCB)
1310.	Fanconi anemia type C (FANCC)

1311.	Fanconi anemia type D1 (BRCA2)
1312.	Fanconi anemia type D2 (FANCD2)
1313.	Fanconi anemia type E (FANCE)
1314.	Fanconi anemia type F (FANCF)
1315.	Fanconi anemia type G (FANCG)
1316.	Fanconi anemia type I (FANCI)
1317.	Fanconi anemia type J (BRIP1)
1318.	Fanconi anemia type L (FANCL)
1319.	Fanconi anemia type M (FANCM)
1320.	Fanconi anemia type N (PALB2)
1321.	Fanconi anemia type P (SLX4)
1322.	Fanconi anemia, complementation group Q (ERCC4)
1323.	Fanconi anemia, XRCCR2 related (XRCC2)
1324.	Fanconi renotubular syndrome type 2 (SLC34A1)
1325.	Fanconi-Bickel syndrome (SLC2A2)
1326.	Farber disease (ASAH1)
1327.	Fatal familial imsomnia (PRNP)
1328.	Favism, susceptibility to (G6PD)
1329.	Fazio-Londe disease (SLC52A3)
1330.	Febrile seizures, familial, type 4 (NGS Sequencing: ADGRV1)
1331.	Feingold syndrome (MYCN)
1332.	Feingold syndrome type 2 (MIR17HG)
1333.	Fetal akinesia deformation sequence (DOK7)
1334.	Fetal akinesia deformation sequence (RAPSN)
1335.	FG syndrome type 1 (MED12)
1336.	FG syndrome type 2 (FLNA)
1337.	FG syndrome type 4 (CASK)
1338.	Fibrochondrogenesis 2 (COL11A2)
1339.	Fibrochondrogenesis type 1 (NGS Sequencing: COL11A1)
1340.	Fibrodysplasia ossificans progressiva (ACVR1)
1341.	Fibrosis of extraocular muscles, congenital type 1 (KIF21A)
1342.	Fibrosis of extraocular muscles, congenital type 2 (PHOX2A)

1343.	Fibrosis of extraocular muscles, congenital type 3a (TUBB3)
1344.	Fibular aplasia or hypoplasia, femoral bowing and poly-, syn-, and oligodactyly (WNT7A)
1345.	Filaminopathy (FLNC)
1346.	FILS syndrome (POLE)
1347.	Fish eye disease (LCAT)
1348.	Fleck retina, familial benign (PLA2G5)
1349.	Floating-Harbor syndrome (SRCAP)
1350.	Focal dermal hypoplasia (PORCN)
1351.	Focal segmental glomerulosclerosis and dilated cardiomyopath, MT-TY related (NGS Sequencing: MT-TY)
1352.	Focal segmental glomerulosclerosis type 1 (ACTN4)
1353.	Focal segmental glomerulosclerosis type 2 (TRPC6)
1354.	Focal segmental glomerulosclerosis type 3 (CD2AP)
1355.	Focal segmental glomerulosclerosis type 4, susceptibility to (APOL1)
1356.	Focal segmental glomerulosclerosis type 5 (INF2)
1357.	Focal segmental glomerulosclerosis type 6 (MYO1E)
1358.	Focal segmental glomerulosclerosis type 7 (PAX2)
1359.	Focal segmental glomerulosclerosis type 8 (ANLN)
1360.	Focal segmental glomerulosclerosis type 9 (CRB2)
1361.	Focal segmental glomerulosclerosis, LAMA5 related (LAMA5)
1362.	Folate malabsorption, hereditary (SLC46A1)
1363.	Follicle-stimulating hormone deficiency, isolated (FSHB)
1364.	Foveal hypoplasia type 1 (PAX6)
1365.	Fragile X syndrome (FMR1)
1366.	Fragile X tremor/ataxia syndrome (FMR1)
1367.	Frank-ter Haar syndrome (SH3PXD2B)
1368.	Fraser syndrome (NGS Sequencing: FRAS1)
1369.	Fraser syndrome (GRIP1)
1370.	Fraser syndrome (FREM2)
1371.	Friedreich ataxia (FXN)
1372.	Frontometaphyseal dysplasia (FLNA)
1373.	Frontonasal dysplasia type 1 (ALX3)
1374.	Frontonasal dysplasia type 2 (ALX4)

1375.	Fructose intolerance (ALDOB)
1376.	Fructose uptake deficiency, SLC2A5 related (SLC2A5)
1377.	Fructose-1,6-bisphosphatase deficiency (FBP1)
1378.	Fructosuria essential (KHK)
1379.	FSH releasing protein deficiency (INHBA)
1380.	Fucosidosis (FUCA1)
1381.	Fukuyama congenital muscular dystrophy (FKTN)
1382.	Fumarase deficiency (FH)
1383.	Fundus albipunctatus (RDH5)
1384.	Fundus albipunctatus (PRPH2)
1385.	GABA-transaminase deficiency (ABAT)
1386.	Galactokinase deficiency (GALK1)
1387.	Galactose epimerase deficiency (GALE)
1388.	Galactosemia (GALT)
1389.	Galactosialidosis (CTSA)
1390.	Gallbladder disease type 1 (ABCB4)
1391.	Galloway-Mowat syndrome (WDR73)
1392.	Gastric cancer, BLM related, somatic (BLM)
1393.	Gastric cancer, hereditary diffuse (CDH1)
1394.	Gastric cancer, somatic (ERBB2)
1395.	Gastric cancer, somatic (FGFR2)
1396.	Gastric cancer, somatic (KLF6)
1397.	Gastric cancer, somatic (KRAS)
1398.	Gastric cancer, somatic (MUTYH)
1399.	Gastric cancer, somatic (APC)
1400.	Gastrointestinal stromal tumor, familial (KIT)
1401.	Gastrointestinal stromal tumor, somatic (PDGFRA)
1402.	Gaucher disease type 1 (GBA)
1403.	Gaucher disease type 2 (GBA)
1404.	Gaucher disease type 3 (GBA)
1405.	Gaucher disease type 3C (GBA)
1406.	Gaucher disease, atypical (PSAP)

1407.	Gaucher disease, perinatal lethal (GBA)
1408.	Gaze palsy, horizontal, with progressive scoliosis (ROBO3)
1409.	Geleophysic dysplasia type 1 (ADAMTSL2)
1410.	Geleophysic dysplasia type 2 (NGS Sequencing: FBN1)
1411.	Generalized epilepsy and paroxysmal dyskinesia (KCNMA1)
1412.	Generalized epilepsy with febrile seizures plus type 1 (SCN1B)
1413.	Generalized epilepsy with febrile seizures plus type 2 (SCN1A)
1414.	Generalized epilepsy with febrile seizures plus type 3 (GABRG2)
1415.	Generalized epilepsy with febrile seizures plus type 7 (SCN9A)
1416.	Genitopatellar syndrome (KAT6B)
1417.	Germ cell tumors, somatic (KIT)
1418.	Geroderma osteodysplasticum (GORAB)
1419.	Gerstmann-Straussler disease (PRNP)
1420.	Ghosal hematodiaphyseal syndrome (TBXAS1)
1421.	Giant axonal neuropathy type 1 (GAN)
1422.	Gilbert syndrome (UGT1A1)
1423.	Gillespie syndrome (NGS Sequencing: ITPR1)
1424.	Gitelman syndrome (SLC12A3)
1425.	Glass syndrome (SATB2)
1426.	Glaucoma, open angle type 1A (MYOC)
1427.	Glaucoma, open angle type 1E (OPTN)
1428.	Glaucoma, open angle type 1F (NTF4)
1429.	Glaucoma, open angle type 1F (ASB10)
1430.	Glaucoma, open angle type 1G (WDR36)
1431.	Glaucoma, primary type 3A (CYP1B1)
1432.	Glaucoma, primary type 3D (LTBP2)
1433.	Glioblastoma type 3, susceptibility to, due to BRCA2 germline mutation (BRCA2)
1434.	Glioblastoma, somatic (ERBB2)
1435.	Glioma susceptibility 1, susceptibility to, somatic (TP53)
1436.	Glioma, susceptibility to, somatic (IDH1)
1437.	Glomerulocystic kidney disease with hyperuricemia and isosthenuria (UMOD)
1438.	Glucocorticoid deficiency type 1 (MC2R)

1439.	Glucocorticoid deficiency type 2 (MRAP)
1440.	Glucocorticoid resistance, generalized (NR3C1)
1441.	Glucose/Galactose malabsorption (SLC5A1)
1442.	GLUT1 deficiency syndrome type 1 (SLC2A1)
1443.	Glutamate formiminotransferase deficiency (FTCD)
1444.	Glutamine deficiency, congenital (GLUL)
1445.	Glutaric acidemia type 1 (GCDH)
1446.	Glutaric acidemia type 2C (ETFDH)
1447.	Glutaric aciduria type 3 (SUGCT)
1448.	Glutathione S-transferase theta-1 defficiency (GSTT1)
1449.	Glutathione synthetase deficiency (GSS)
1450.	Glycerol kinase deficiency (GK)
1451.	Glycine encephalopathy (AMT)
1452.	Glycine encephalopathy (GCSH)
1453.	Glycine encephalopathy (GLDC)
1454.	Glycine N-methyltransferase deficiency (GNMT)
1455.	Glycogen storage disease of heart (lethal) (PRKAG2)
1456.	Glycogen storage disease type 0 (GYS2)
1457.	Glycogen storage disease type 0 muscle (GYS1)
1458.	Glycogen storage disease type 10 (PGAM2)
1459.	Glycogen storage disease type 11 (LDHA)
1460.	Glycogen storage disease type 12 (ALDOA)
1461.	Glycogen storage disease type 13 (ENO3)
1462.	Glycogen storage disease type 14 (PGM1)
1463.	Glycogen storage disease type 15 (GYG1)
1464.	Glycogen storage disease type 1A (G6PC)
1465.	Glycogen storage disease type 1B (SLC37A4)
1466.	Glycogen storage disease type 1C (SLC37A4)
1467.	Glycogen storage disease type 2 (GAA)
1468.	Glycogen storage disease type 3 (AGL)
1469.	Glycogen storage disease type 4 (GBE1)
1470.	Glycogen storage disease type 5 (PYGM)

1471.	Glycogen storage disease type 6B (PYGL)
1472.	Glycogen storage disease type 7 (PFKM)
1473.	Glycogen storage disease type 9A (PHKA2)
1474.	Glycogen storage disease type 9B (PHKB)
1475.	Glycogen storage disease type 9C (PHKG2)
1476.	Glycoprotein Ia C807T polymorphism (ITGA2)
1477.	Glycosylation disorde type 2A (MGAT2)
1478.	Glycosylation disorde type 2C (SLC35C1)
1479.	Glycosylation disorder type 1A (PMM2)
1480.	Glycosylation disorder type 1B (MPI)
1481.	Glycosylation disorder type 1C (ALG6)
1482.	Glycosylation disorder type 1D (ALG3)
1483.	Glycosylation disorder type 1E (DPM1)
1484.	Glycosylation disorder type 1F (MPDU1)
1485.	Glycosylation disorder type 1G (ALG12)
1486.	Glycosylation disorder type 1H (ALG8)
1487.	Glycosylation disorder type 1I (ALG2)
1488.	Glycosylation disorder type 1J (DPAGT1)
1489.	Glycosylation disorder type 1K (ALG1)
1490.	Glycosylation disorder type 1L (ALG9)
1491.	Glycosylation disorder type 1M (DOLK)
1492.	Glycosylation disorder type 1N (RFT1)
1493.	Glycosylation disorder type 1O (DPM3)
1494.	Glycosylation disorder type 1S (ALG13)
1495.	Glycosylation disorder type 1U (DPM2)
1496.	Glycosylation disorder type 2A (MGAT2)
1497.	Glycosylation disorder type 2B (MOGS)
1498.	Glycosylation disorder type 2C (SLC35C1)
1499.	Glycosylation disorder type 2D (B4GALT1)
1500.	Glycosylation disorder type 2E (COG7)
1501.	Glycosylation disorder type 2F (SLC35A1)
1502.	Glycosylation disorder type 2G (COG1)

1503.	Glycosylation disorder type 2H (COG8)
1504.	Glycosylation disorder type 2I (COG5)
1505.	Glycosylation disorder type 2J (COG4)
1506.	Glycosylation disorder type 2K (TMEM165)
1507.	Glycosylation disorder type 2M (SLC35A2)
1508.	Glycosylation disorder type 3 (COG6)
1509.	Glycosylation disorder type IR (DDOST)
1510.	Glycosylation disorder x-linked (SSR4)
1511.	GM1-gangliosidosis (GLB1)
1512.	GM1-gangliosidosis type 1 (GLB1)
1513.	GM1-gangliosidosis type 2 (GLB1)
1514.	GM2-gangliosidosis type 2 (HEXB)
1515.	Gnathodiaphyseal dysplasia (ANO5)
1516.	Goitre, multinodular (KEAP1)
1517.	Goldberg-Shprintzen megacolon syndrome (KIF1BP)
1518.	Gracile bone dysplasia (FAM111A)
1519.	GRACILE syndrome (BCS1L)
1520.	Granulomatous disease, chronic, autosomal recessive, cytochrome b- positive, type 1 (NCF1)
1521.	Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative (CYBA)
1522.	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 2 (NCF2)
1523.	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 3 (NCF4)
1524.	Granulomatous disease, chronic, X-linked (CYBB)
1525.	Greenberg skeletal dysplasia (LBR)
1526.	Greig cephalopolysyndactyly syndrome (GLI3)
1527.	Griscelli syndrome type 1 (MYO5A)
1528.	Griscelli syndrome type 2 (RAB27A)
1529.	Griscelli syndrome type 3 (MLPH)
1530.	Growth hormone deficiency (GH1)
1531.	Growth hormone deficiency (GHRHR)
1532.	Growth hormone insensitivity with immunodeficiency (STAT5B)
1533.	Growth hormone insensitivity, partial (GHR)
1534.	Growth retardation with deafness and mental retardation due to IGF1 deficiency (IGF1)

1535.	Guanidinoacetate methyltransferase deficiency (GAMT)
1536.	Guttmacher syndrome (HOXA13)
1537.	Gyrate atrophy of choroid and retina with or without ornithinemia (OAT)
1538.	Haim-Munk syndrome (CTSC)
1539.	Hamamy syndrome (IRX5)
1540.	Hand-foot-uterus syndrome (HOXA13)
1541.	Hartnup disorder (SLC6A19)
1542.	Hartsfield syndrome (FGFR1)
1543.	Hawkinsinuria (HPD)
1544.	HDL deficiency, type 2 (ABCA1)
1545.	Hearing loss, MAP1A related (MAP1A)
1546.	Hearing loss, MYH7B related (MYH7B)
1547.	Heart block, progressive, familial, type 1A (SCN5A)
1548.	Heart-hand syndrome, Slovenian type (LMNA)
1549.	Heimler syndrome type 1 (PEX1)
1550.	Helsmoortel-van der Aa syndrome (ADNP)
1551.	Hemangioblastoma, cerebellar, somatic (VHL)
1552.	Hemangioma capillary infantile (ANTXR1)
1553.	Hemangioma, capillary infantile, familial, susceptibility to (KDR)
1554.	Hemochromatosis classical (HFE)
1555.	Hemochromatosis type 2A (HFE2)
1556.	Hemochromatosis type 2B (HAMP)
1557.	Hemochromatosis type 3 (TFR2)
1558.	Hemochromatosis type 4 (SLC40A1)
1559.	Hemolytic anemia due to G6PD deficiency (G6PD)
1560.	Hemolytic anemia due to triosephosphate isomerase deficiency (TPI1)
1561.	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy (CD59)
1562.	Hemolytic anemia, Kell-system related (KEL)
1563.	Hemolytic uremic syndrome (CFH)
1564.	Hemolytic uremic syndrome (CFI)
1565.	Hemolytic uremic syndrome (THBD)
1566.	Hemolytic uremic syndrome (CFB)

1567.	Hemolytic uremic syndrome (CFHR1)
1568.	Hemolytic uremic syndrome (CFHR2)
1569.	Hemolytic uremic syndrome (CFHR3)
1570.	Hemolytic uremic syndrome (CFHR4)
1571.	Hemolytic uremic syndrome (CFHR5)
1572.	Hemolytic uremic syndrome, atypical type 2, susceptibility to (CD46)
1573.	Hemophagocytic lymphohistiocytosis type 2 (PRF1)
1574.	Hemophagocytic lymphohistiocytosis type 3 (UNC13D)
1575.	Hemophagocytic lymphohistiocytosis type 4 (STX11)
1576.	Hemophagocytic lymphohistiocytosis type 5 (STXBP2)
1577.	Hemophilia A (F8)
1578.	Hemophilia B (F9)
1579.	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts (JAM3)
1580.	Hennekam lymphangiectasia-lymphedema syndrome type 1 (CCBE1)
1581.	Hennekam lymphangiectasia-lymphedema syndrome type 2 (FAT4)
1582.	Hepatic failure, early onset, and neurologic disorder (SCO1)
1583.	Hepatic venoocclusive disease with immunodeficiency (SP110)
1584.	Hepatoblastoma, somatic (APC)
1585.	Hepatocellular carcinoma, somatic (CTNNB1)
1586.	Hepatocellular carcinoma, somatic (IGF2R)
1587.	Hepatocellular carcinoma, somatic (TP53)
1588.	Hereditary breast and ovarian cancer syndrome, RAD50 related (RAD50)
1589.	Hereditary motor and sensory neuropathy, Okinawa type (TFG)
1590.	Hereditary myopathy with early respiratory failure (NGS Sequencing: TTN)
1591.	Hereditary persistence of fetal hemoglobin (HBG2)
1592.	Hereditary Retinoblastoma (RB1)
1593.	Hermansky Pudlak syndrome type 4 (HPS4)
1594.	Hermansky-Pudlak syndrome type 1 (HPS1)
1595.	Hermansky-Pudlak syndrome type 2 (AP3B1)
1596.	Hermansky-Pudlak syndrome type 3 (HPS3)
1597.	Hermansky-Pudlak syndrome type 5 (HPS5)
1598.	Hermansky-Pudlak syndrome type 6 (HPS6)

1599.	Hermansky-Pudlak syndrome type 7 (DTNBP1)
1600.	Hermansky-Pudlak syndrome type 8 (BLOC1S3)
1601.	Herpes simplex encephalitis type 2, susceptibility to (TLR3)
1602.	Heterotaxy, visceral type 1 (ZIC3)
1603.	Heterotaxy, visceral type 2 (CFC1)
1604.	Heterotaxy, visceral type 4 (ACVR2B)
1605.	Heterotaxy, visceral type 5 (NODAL)
1606.	Heterotaxy, visceral type 6 (CFAP53)
1607.	Heterotaxy, visceral type 7 (MMP21)
1608.	Heterotaxy, visceral, BCL9L related (BCL9L)
1609.	Heterotopia, periventricular, ED variant (FLNA)
1610.	Heterotopia, periventricular, X-linked dominant (FLNA)
1611.	Hippocampal longterm potentiation, RFN39 related (RNF39)
1612.	Hirschsprung disease (KIF1BP)
1613.	Hirschsprung disease (EDNRB)
1614.	Hirschsprung disease (EDN3)
1615.	Hirschsprung disease (ECE1)
1616.	Hirschsprung disease (ZEB2)
1617.	Hirschsprung disease (NRTN)
1618.	Hirschsprung disease (NRG1)
1619.	Hirschsprung disease (RET)
1620.	Hirschsprung disease, type 3, susceptibility to (GDNF)
1621.	Histiocytosis-lymphadenopathy plus syndrome (SLC29A3)
1622.	Holocarboxylase synthetase deficiency (HLCS)
1623.	Holoprosencephaly type 11 (CDON)
1624.	Holoprosencephaly type 2 (SIX3)
1625.	Holoprosencephaly type 3 (SHH)
1626.	Holoprosencephaly type 4 (TGIF1)
1627.	Holoprosencephaly type 5 (ZIC2)
1628.	Holoprosencephaly-type 9 (GLI2)
1629.	Holt-Oram syndrome (TBX5)
1630.	Homocystinuria (MTHFR)

1631.	Homocystinuria due to cystathionine beta-synthase deficiency (CBS)
1632.	Homocystinuria-megaloblastic anemia, cbl E type (MTRR)
1633.	Hoyeraal-Hreidarsson syndrome (DKC1)
1634.	HSAN1 (SPTLC1)
1635.	HSAN2A (WNK1)
1636.	HSAN2B (FAM134B)
1637.	HSAN3 (IKBKAP)
1638.	HSAN4 (NTRK1)
1639.	HSAN5 (NGF)
1640.	HSAN8 (PRDM12)
1641.	HSN2C (KIF1A)
1642.	Huntington disease (HTT)
1643.	Huntington disease, ZDHHC17 related (ZDHHC17)
1644.	Huntington disease-like type 1 (PRNP)
1645.	Huntington disease-like type 2 (JPH3)
1646.	Hurler syndrome (IDUA)
1647.	Hurler-Scheie syndrome (IDUA)
1648.	Hurthle cell thyroid carcinoma, due to germline NDUFA13 mutation (NDUFA13)
1649.	Hutchinson-Gilford progeria (LMNA)
1650.	Hyaline fibromatosis syndrome (ANTXR2)
1651.	Hydatidiform mole (NLRP7)
1652.	Hydatidiform mole, recurrent, type 2 (KHDC3L)
1653.	Hydranencephaly with abnormal genitalia/Lissencephaly X-linked 2 (ARX)
1654.	Hydranencephaly, Fowler type (FLVCR2)
1655.	Hydrocephalus with aqueductal stenosis and congenital intestinal pseudoobstraction (L1CAM)
1656.	Hydrocephalus, nonsyndromic, autosomal recessive type 1 (CCDC88C)
1657.	Hydrocephalus, nonsyndromic, autosomal recessive type 2 (MPDZ)
1658.	Hydrolethalus syndrome (HYLS1)
1659.	Hyper-IgE recurrent infection syndrome (STAT3)
1660.	Hyper-IgE recurrent infection syndrome, autosomal recessive (DOCK8)
1661.	Hyperaldosteronism type 3 (KCNJ5)
1662.	Hyperbilirubinemia, familial transient neonatal (UGT1A1)

1663.	Hyperbilirubinemia, Rotor type (SLCO1B3)
1664.	Hyperbilirubinemia, Rotor type (SLCO1B1)
1665.	Hypercalcemia infantile type (CYP24A1)
1666.	Hypercholanemia (BAAT)
1667.	Hypercholanemia (TJP2)
1668.	Hypercholesterolemia autosomal dominant type 3 (PCSK9)
1669.	Hypercholesterolemia autosomal recessive (LDLRAP1)
1670.	Hypercholesterolemia due to LDL-receptor-disorder autosomal dominant (LDLR)
1671.	Hypercholesterolemia type B autosomanl dominant (APOB)
1672.	Hyperchylomicronemia type 5 (APOA5)
1673.	Hyperekplexia (SLC6A5)
1674.	Hyperekplexia (GLRB)
1675.	Hyperekplexia (GLRA1)
1676.	Hyperekplexia, EIEE8 related (ARHGEF9)
1677.	Hypereosinophilic syndrome, idiopathic, resistant to imatinib (PDGFRA)
1678.	Hyperferritinemia-cataract syndrome (FTL)
1679.	Hyperinsulinaemia, association with, G6PC2 related (G6PC2)
1680.	Hyperinsulinemic hypoglycemia type 1 (ABCC8)
1681.	Hyperinsulinemic hypoglycemia type 2 (KCNJ11)
1682.	Hyperinsulinemic hypoglycemia type 3 (GCK)
1683.	Hyperinsulinemic hypoglycemia type 6 (GLUD1)
1684.	Hyperinsulinemic hypoglycemia type 7 (SLC16A1)
1685.	Hyperinsulinism, UCP2 related (UCP2)
1686.	Hyperkalemic periodic paralysis (SCN4A)
1687.	Hyperlipidemia, familial combined, susceptibility to (USF1)
1688.	Hyperlipoproteinemia type 1 (LPL)
1689.	Hyperlysinemia type 1 (AASS)
1690.	Hypermanganesemia with dystonia, polycythemia and cirrhosis (SLC30A10)
1691.	Hypermethioninemia due to adenosine kinase deficiency (ADK)
1692.	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase (AHCY)
1693.	Hyperornithinemia- Hyperammonemia - Homocitrullinuria syndrome (SLC25A15)
1694.	Hyperoxaluria type 1 (AGXT)

1695.	Hyperoxaluria type 2 (GRHPR)
1696.	Hyperoxaluria type 3 (HOGA1)
1697.	Hyperoxaluria, SLC26A6 related (SLC26A6)
1698.	Hyperparathyroidism type 1, familial (CDC73)
1699.	Hyperparathyroidism type 2, familial (CDC73)
1700.	Hyperparathyroidism, neonatal severe (CASR)
1701.	Hyperphenylalaninemia, BH4 deficient, type C (QDPR)
1702.	Hyperphenylalaninemia, BH4 deficient, type D (PCBD1)
1703.	Hyperphenylalaninemia, BH4-deficient, B (GCH1)
1704.	Hyperphenylalaninemia, BH4-deficient, type A (PTS)
1705.	Hyperphosphatasia with mental retardation syndrome type 1 (PIGV)
1706.	Hyperphosphatasia with mental retardation syndrome type 2 (PIGO)
1707.	Hyperphosphatasia with mental retardation syndrome type 3 (PGAP2)
1708.	Hyperphosphatasia with mental retardation syndrome type 4 (PGAP3)
1709.	Hyperphosphatasia with mental retardation syndrome type 5 (PIGW)
1710.	Hyperprolinemia type 1 (PRODH)
1711.	Hypertension early onset (NR3C2)
1712.	Hypertension, ADD2 related (ADD2)
1713.	Hypertension, salt-sensitive essential, susceptibility to (CYP3A5)
1714.	Hypertriglyceridemia, susceptibility to (LIPI)
1715.	Hypertriglyceridemia, transient infantile (GPD1)
1716.	Hypertrophic osteoarthropathy type 1 (HPGD)
1717.	Hypertrophic osteoarthropathy type 2 (SLCO2A1)
1718.	Hyperuricemic nephropathy, familial juvenile type 1 (UMOD)
1719.	Hypoaldosteronism congenital due to CMO I deficiency (CYP11B2)
1720.	Hypoaldosteronism, congenital, due to CMO II deficiency (CYP11B2)
1721.	Hypoalphalipoproteinemia (APOA1)
1722.	Hypobetalipoproteinemia type 1 (APOB)
1723.	Hypocalcemia, autosomal dominant 2 (GNA11)
1724.	Hypocalcemia, autosomal dominant, with Bartter syndrome (CASR)
1725.	Hypocalciuric hypercalcemia, familial type 3 (AP2S1)
1726.	Hypocalciuric hypercalcemia, type 1 (CASR)

1727.	Hypochondroplasia (FGFR3)
1728.	Hypoglycemia of infancy, leucine-sensitive (ABCC8)
1729.	Hypogonadism, alopecia, Diabetes mellitus, mental retardation and extrapyramidal syndrome (DCAF17)
1730.	Hypogonadotropic hypogonadism (KISS1)
1731.	Hypogonadotropic hypogonadism (NSMF)
1732.	Hypogonadotropic hypogonadism (LHB)
1733.	Hypogonadotropic hypogonadism (KISS1R)
1734.	Hypogonadotropic hypogonadism type 10 with or without anosmia (TAC3)
1735.	Hypogonadotropic hypogonadism type 11 with or without anosmia (TACR3)
1736.	Hypogonadotropic hypogonadism type 12 with or without anosmia (GNRH1)
1737.	Hypogonadotropic hypogonadism type 15 with or without anosmia (HS6ST1)
1738.	Hypogonadotropic hypogonadism type 3 with or without anosmia (PROKR2)
1739.	Hypogonadotropic hypogonadism type 6 with or without anosmia (FGF8)
1740.	Hypogonadotropic hypogonadism type 7 with or without anosmia (GNRHR)
1741.	Hypogonadtropic hypogonadism type 14 (WDR11)
1742.	Hypoinsulinemic hypoglycemia with hemihypertrophy (AKT2)
1743.	Hypokalemic periodic paralysis type 1 (CACNA1S)
1744.	Hypomagnesemia type 1 (TRPM6)
1745.	Hypomagnesemia type 2 (FXYD2)
1746.	Hypomagnesemia type 3 (CLDN16)
1747.	Hypomagnesemia type 4 (EGF)
1748.	Hypomagnesemia type 5 (CLDN19)
1749.	Hypomagnesemia type 6 (CNNM2)
1750.	Hypomyelination with brainstem and spinal cord involvement and leg spasticity (DARS)
1751.	Hypoparathyroidism (PTH)
1752.	Hypoparathyroidism, familial isolated (GCM2)
1753.	Hypoparathyroidism, sensorineural deafness, and renal dysplasia (GATA3)
1754.	Hypoparathyroidism-retardation-dysmorphism syndrome (TBCE)
1755.	Hypophosphatasia, adult (ALPL)
1756.	Hypophosphatasia, childhood (ALPL)
1757.	Hypophosphatasia, infantile (ALPL)
1758.	Hypophosphatemic rickets (CLCN5)

1759.	Hypophosphatemic rickets with hypercalciuria (SLC34A3)
1760.	Hypophosphatemic rickets, autosomal dominant (FGF23)
1761.	Hypophosphatemic rickets, autosomal recessive type 1 (DMP1)
1762.	Hypophosphatemic rickets, autosomal recessive type 2 (ENPP1)
1763.	Hypophosphatemic rickets, X-linked (PHEX)
1764.	Hypospadias type 1, X-linked (AR)
1765.	Hypospadias type 2, X-linked (MAMLD1)
1766.	Hypothyroidism congenital nongoitrous type 1 (TSHR)
1767.	Hypothyroidism congenital nongoitrous type 2, familial (PAX8)
1768.	Hypothyroidism congenital nongoitrous type 4 (TSHB)
1769.	Hypothyroidism congenital nongoitrous type 6 (THRA)
1770.	Hypothyroidism, isolated, TRHR related (TRHR)
1771.	Hypotonia-cystinuria syndrome (PREPL)
1772.	Hypotrichosis type 1 (APCDD1)
1773.	Hypotrichosis type 11 (SNRPE)
1774.	Hypotrichosis type 12 (RPL21)
1775.	Hypotrichosis type 13 (KRT71)
1776.	Hypotrichosis type 2 (CDSN)
1777.	Hypotrichosis type 3 (KRT74)
1778.	Hypotrichosis type 4 (HR)
1779.	Hypotrichosis type 6 (DSG4)
1780.	Hypotrichosis type 7 (LIPH)
1781.	Hypotrichosis type 8 (LPAR6)
1782.	Hypotrichosis-lymphedema-telangiectasia syndrome (SOX18)
1783.	Hypouricemia, renal type 1 (SLC22A12)
1784.	Hypouricemia, renal type 2 (SLC2A9)
1785.	Ichthyosiform erythroderma, congenital, nonbullous type 1 (ALOXE3)
1786.	Ichthyosiform erythroderma, congenital, nonbullous type 1 (NIPAL4)
1787.	Ichthyosis congenital, autosomal recessive, PNPLA1 related (PNPLA1)
1788.	Ichthyosis congenital, Harlequin fetus type (ABCA12)
1789.	Ichthyosis follicularis, atricia, and photophobia syndrome (MBTPS2)
1790.	Ichthyosis prematurity syndrome (SLC27A4)

1791.	Ichthyosis vulgaris (FLG)
1792.	Ichthyosis, bullous type (KRT2)
1793.	Ichthyosis, congenital, autosomal recessive type 1 (TGM1)
1794.	Ichthyosis, congenital, autosomal recessive, type 11 (ST14)
1795.	Ichthyosis, congenital, autosomal recessive, type 2 (ALOX12B)
1796.	Ichthyosis, congenital, autosomal recessive, type 9 (CERS3)
1797.	Ichthyosis, lamellar type 2 (ABCA12)
1798.	Ichthyosis, lamellar type 3 (CYP4F22)
1799.	Ichthyosis, lamellar type 4 (LIPN)
1800.	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis (CLDN1)
1801.	Ichthyosis, spastic quadriplegia, and mental retardation (ELOVL4)
1802.	Ichthyosis, X-linked (STS)
1803.	IDH1, selective sequencing of exon 4 (IDH1)
1804.	IDH2, selective sequencing of exon 4 (IDH2)
1805.	IMAGE syndrome (CDKN1C)
1806.	Immunodeficiency common variable type 1 (ICOS)
1807.	Immunodeficiency common variable type 10 (NFKB2)
1808.	Immunodeficiency common variable type 2 (TNFRSF13B)
1809.	Immunodeficiency common variable type 3 (CD19)
1810.	Immunodeficiency common variable type 4 (TNFRSF13C)
1811.	Immunodeficiency common variable type 6 (CD81)
1812.	Immunodeficiency common variable type 8 (LRBA)
1813.	Immunodeficiency due to defect in MAPBP-interacting protein (LAMTOR2)
1814.	Immunodeficiency due to purine nucleoside phosphorylase deficiency (PNP)
1815.	Immunodeficiency type 10 (STIM1)
1816.	Immunodeficiency type 11 (CARD11)
1817.	Immunodeficiency type 12 (MALT1)
1818.	Immunodeficiency type 14 (PIK3CD)
1819.	Immunodeficiency type 15 (IKBKB)
1820.	Immunodeficiency type 18 (CD3E)
1821.	Immunodeficiency type 19 (CD3D)
1822.	Immunodeficiency type 2, with hyper-IgM (AICDA)

1823.	Immunodeficiency type 21 (GATA2)
1824.	Immunodeficiency type 22 (LCK)
1825.	Immunodeficiency type 24 (CTPS1)
1826.	Immunodeficiency type 25 (CD247)
1827.	Immunodeficiency type 26, with or without neurologic abnormalities (NGS Sequencing: PRKDC)
1828.	Immunodeficiency type 3, with hyper-IgM (CD40)
1829.	Immunodeficiency type 32A, mycobacteriosis, autosomal dominant (IRF8)
1830.	Immunodeficiency type 32B, monocyte and dendritic cell deficiency, autosomal recessive (IRF8)
1831.	Immunodeficiency type 34 (CYBB)
1832.	Immunodeficiency type 35 (TYK2)
1833.	Immunodeficiency type 36 (PIK3R1)
1834.	Immunodeficiency type 38 (ISG15)
1835.	Immunodeficiency type 5, with hyper IgM (UNG)
1836.	Immunodeficiency type 8 (CORO1A)
1837.	Immunodeficiency type 9 (ORAI1)
1838.	Immunodeficiency with natural killer cell deficiency (MCM4)
1839.	Immunodeficiency, common variable type 7 (CR2)
1840.	Immunodeficiency, isolated (IKBKG)
1841.	Immunodeficiency, primary, autosomal recessive, IL21R-related (IL21R)
1842.	Immunodeficiency, X-linked with hyper-IgM (CD40LG)
1843.	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia (MAGT1)
1844.	Immunodeficiency-centromeric instability-facial anomalies syndrome type 1 (DNMT3B)
1845.	Immunodeficiency-centromeric instability-facial anomalies syndrome type 2 (ZBTB24)
1846.	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked (FOXP3)
1847.	Immunological disorder, PECAM1 related (PECAM1)
1848.	Immunological disorder, PICALM related (PICALM)
1849.	Inclusion body myopathy (GNE)
1850.	Inclusion body myopathy (MYH2)
1851.	Incontinentia pigmenti type 2 (IKBKG)
1852.	Infantile liver failure syndrome type 2 (NBAS)
1853.	Infantile neuroaxonal dystrophy type 1 (PLA2G6)
1854.	Inflammatory bowel disease type 13 (ABCB1)

1855.	Inflammatory skin and bowel disease, neonatal, type 1 (ADAM17)
1856.	Insensitivity to pain, channelopathy-associated (SCN9A)
1857.	Insulin-like growth factor resistance (IGF1R)
1858.	Intellectual disability nonsyndromic, CIC related (CIC)
1859.	Intellectual disability nonsyndromic, CNKSR2 related (CNKSR2)
1860.	Intellectual disability, TBR1 related (TBR1)
1861.	Interleukin 12A deficiency (IL12A)
1862.	Interleukin 2 receptor deficiency (IL2RA)
1863.	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital (ITGA3)
1864.	Interstitial nephritis karyomegalic (FAN1)
1865.	Intestinal atresia, multiple (TTC7A)
1866.	Intestinal pseudoobstraction, neuronal (FLNA)
1867.	Intrahepatic cholestasis of pregnancy, NR1H4 related (NR1H4)
1868.	Intrinsic factor deficiency (GIF)
1869.	Invasive pneumococcal disease, recurrent isolated type 1 (IRAK4)
1870.	Invasive pneumococcal disease, recurrent isolated type 2 (IKBKG)
1871.	IRAK4 deficiency (IRAK4)
1872.	Iridogoniodysgenesis type 1 (FOXC1)
1873.	Iron-refractory iron deficiency anemia (TMPRSS6)
1874.	Isobutyryl-CoA dehydrogenase deficiency (ACAD8)
1875.	Isovaleric acidemia (IVD)
1876.	IVIC syndrome (SALL4)
1877.	Jackson-Weiss syndrome (FGFR1)
1878.	Jackson-Weiss syndrome (FGFR2)
1879.	JAK2, selective sequencing of exons 12, 14 and 16 (JAK2)
1880.	Jalili syndrome (CNNM4)
1881.	Jawad syndrome (RBBP8)
1882.	Jensen syndrome (TIMM8A)
1883.	Jervell and Lange-Nielsen syndrome type 1 (KCNQ1)
1884.	Jervell and Lange-Nielsen syndrome type 2 (KCNE1)
1885.	Johanson Blizzard syndrome (UBR1)
1886.	Joubert syndrome type 1 (INPP5E)

1887.	Joubert syndrome type 10 (OFD1)
1888.	Joubert syndrome type 13 (TCTN1)
1889.	Joubert syndrome type 14 (TMEM237)
1890.	Joubert syndrome type 15 (CEP41)
1891.	Joubert syndrome type 16 (TMEM138)
1892.	Joubert syndrome type 17 (C5orf42)
1893.	Joubert syndrome type 18 (TCTN3)
1894.	Joubert syndrome type 2 (TMEM216)
1895.	Joubert syndrome type 20 (TMEM231)
1896.	Joubert syndrome type 21 (CSPP1)
1897.	Joubert syndrome type 22 (PDE6D)
1898.	Joubert syndrome type 23 (KIAA0586)
1899.	Joubert syndrome type 24 (TCTN2)
1900.	Joubert syndrome type 3 (AHI1)
1901.	Joubert syndrome type 4 (NPHP1)
1902.	Joubert syndrome type 5 (CEP290)
1903.	Joubert syndrome type 6 (TMEM67)
1904.	Joubert syndrome type 7 (RPGRIP1L)
1905.	Joubert syndrome type 8 (ARL13B)
1906.	Joubert syndrome type 9 (CC2D2A)
1907.	Joubert syndrome, EXOC8 related (EXOC8)
1908.	Joubert syndrome, EXOSC8 related (EXOSC8)
1909.	Juvenile myelomonocytic leukemia, due to CBL germline mutation (CBL)
1910.	Juvenile polyposis syndrome (SMAD4)
1911.	Juvenile polyposis syndrome (BMPR1A)
1912.	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome (SMAD4)
1913.	Kabuki syndrome type 1 (KMT2D)
1914.	Kabuki syndrome type 2 (KDM6A)
1915.	Kagami-Ogata syndrome (paternal UPD chr. 14)
1916.	Kallmann syndrome type 1 (ANOS1)
1917.	Kallmann syndrome type 2 (FGFR1)
1918.	Kallmann syndrome type 4 (PROK2)

1919.	Kallmann syndrome type 5 (CHD7)
1920.	Kallmann syndrome, SEMA3A related (SEMA3A)
1921.	KBG syndrome (ANKRD11)
1922.	Kenny-Caffey syndrome type 2 (FAM111A)
1923.	Keratitis ichthyosis deafness syndrome autosomal dominant (GJB2)
1924.	Keratoconus type 1 (VSX1)
1925.	Keratoderma, palmoplantar, punctate type 1A (AAGAB)
1926.	Keratoderma, palmoplantar, with deafness (GJB2)
1927.	Keratosis follicularis spinulosa declavans, X-linked (MBTPS2)
1928.	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma (POMP)
1929.	Keratosis palmoplantaris striata type 1 (DSG1)
1930.	Keratosis palmoplantaris striata type 2 (DSP)
1931.	Keutel syndrome (MGP)
1932.	Kindler syndrome (FBLIM1)
1933.	Kindler syndrome (FERMT1)
1934.	King-Denborough syndrome (NGS Sequencing: RYR1)
1935.	KIT, selective sequencing of exons 8, 9, 11, 13 and 17 (KIT)
1936.	Kleefstra syndrome (EHMT1)
1937.	Klippel-Feil syndrome type 1, autosomal dominant (GDF6)
1938.	Klippel-Feil syndrome type 2, autosomal dominant (MEOX1)
1939.	Klippel-Feil syndrome type 3, autosomal dominant (GDF3)
1940.	Klippel-Feil syndrome type 4, autosomal dominant, with myopathy and facial dysmorphism (MYO18B)
1941.	Kniest dysplasia (COL2A1)
1942.	Knobloch syndrome type 1 (COL18A1)
1943.	Knuckle pads and leukonychia sensorineural deafness (GJB2)
1944.	Kohlschutter Tonz syndrome (ROGDI)
1945.	Koolen syndrome (KANSL1)
1946.	Krabbe disease (GALC)
1947.	Krabbe disease, atypical (PSAP)
1948.	KRAS somatic Hotspot: c.35G>A, p.G12D (KRAS)
1949.	KRAS somatic Hotspot: c.35G>T p.G12V (KRAS)
1950.	KRAS somatic Hotspot: c.38G>A, p.G13D (KRAS)

1951.	KRAS, selective sequencing of exon 2 and 3 (KRAS)
1952.	L-2-hydroxyglutaric aciduria (L2HGDH)
1953.	Lactase deficiency, congenital (LCT)
1954.	Lactate dehydrogenase-B deficiency (LDHB)
1955.	Lacticacidemia due to PDX1 deficiency (PDHX)
1956.	Lactose intolerance, adult type (MCM6)
1957.	LADD syndrome (FGF10)
1958.	LADD syndrome (FGFR2)
1959.	Langer-Giedion syndrome (EXT1)
1960.	Langer-Giedion syndrome (TRPS1)
1961.	Laron syndrome (GHR)
1962.	Larsen syndrome (FLNB)
1963.	Laryngoonychocutaneous syndrome (NGS Sequencing: LAMA3)
1964.	LCAD deficiency (ACADL)
1965.	LCAT DEFICIENCY (LCAT)
1966.	Leber congenital amaurosis type 1 (GUCY2D)
1967.	Leber congenital amaurosis type 10 (CEP290)
1968.	Leber congenital amaurosis type 11 (IMPDH1)
1969.	Leber congenital amaurosis type 12 (RD3)
1970.	Leber congenital amaurosis type 16 (KCNJ13)
1971.	Leber congenital amaurosis type 17 (GDF6)
1972.	Leber congenital amaurosis type 3 (SPATA7)
1973.	Leber congenital amaurosis type 5 (LCA5)
1974.	Leber congenital amaurosis type 6 (RPGRIP1)
1975.	Leber congenital amaurosis type 7 (CRX)
1976.	Leber congenital amaurosis type 8 (CRB1)
1977.	Leber congenital amaurosis type 9 (NMNAT1)
1978.	Leber congenital amaurosis with myopathy (DTHD1)
1979.	Leber optic atrophy (NGS Sequencing: MT-ND6)
1980.	Leber optic atrophy (NGS Sequencing: MT-ND5)
1981.	Leber optic atrophy (NGS Sequencing: MT-CO1)
1982.	Leber optic atrophy (NGS Sequencing: MT-CO3)

1983.	Leber optic atrophy (NGS Sequencing: MT-ND4L)
1984.	Leber optic atrophy (NGS Sequencing: MT-ND4)
1985.	Leber optic atrophy (NGS Sequencing: MT-ND2)
1986.	Leber optic atrophy (NGS Sequencing: MT-ATP6)
1987.	Leber optic atrophy (NGS Sequencing: MT-CYB)
1988.	Leber optic atrophy (NGS Sequencing: MT-ND1)
1989.	Left-right axis malformations (LEFTY2)
1990.	Legg-Calve-Perthes disease (COL2A1)
1991.	Legius syndrome (SPRED1)
1992.	Leigh syndrome (NDUFA10)
1993.	Leigh syndrome (NDUFAF3)
1994.	Leigh syndrome (NDUFAF1)
1995.	Leigh syndrome (NDUFAF2)
1996.	Leigh syndrome (NDUFS3)
1997.	Leigh syndrome (NDUFA9)
1998.	Leigh syndrome (NDUFS7)
1999.	Leigh syndrome (NDUFS4)
2000.	Leigh syndrome (NDUFS8)
2001.	Leigh syndrome (BCS1L)
2002.	Leigh syndrome (NDUFAF6)
2003.	Leigh syndrome (SDHA)
2004.	Leigh syndrome (FOXRED1)
2005.	Leigh syndrome (NUBPL)
2006.	Leigh syndrome (COX15)
2007.	Leigh syndrome (NDUFA2)
2008.	Leigh syndrome and mitochondrial encephalopathy (ACAD9)
2009.	Leigh syndrome due to COX deficiency (SURF1)
2010.	Leigh syndrome due to mitochondrial complex I deficiency (NGS Sequencing: MT-ND3)
2011.	Leigh syndrome due to mitochondrial complex I deficiency (NDUFA12)
2012.	Leigh syndrome due to mitochondrial complex I deficiency (NGS Sequencing: MT-ND6)
2013.	Leigh syndrome due to mitochondrial complex I deficiency (NGS Sequencing: MT-ND5)
2014.	Leigh syndrome due to pyruvate and alpha-ketoglutarate dehydrogenase deficiencies, LIPT1 related (LIPT1)

2015.	Leigh syndrome due to pyruvate carboxylase deficiency (PC)
2016.	Leigh syndrome due to the mitochondrial complex IV deficiency (TACO1)
2017.	Leigh syndrome, French-Canadian type (LRPPRC)
2018.	Leigh syndrome, X-linked (PDHA1)
2019.	Lenz-Majewski hyperostotic dwarfism (PTDSS1)
2020.	LEOPARD syndrome type 3 (BRAF)
2021.	Lesch-Nyham syndrome (HPRT1)
2022.	Lethal congenital contracture syndrome type 1 (GLE1)
2023.	Lethal congenital contracture syndrome type 4 (MYBPC1)
2024.	Leukemia, acute lymphoblastic (IKZF1)
2025.	Leukemia, acute lymphoblastic, susceptibility to (PAX5)
2026.	Leukemia, acute lymphoblastic, susceptibility to, due to PAX5 germline mutation (PAX5)
2027.	Leukemia, acute myelogenous (KRAS)
2028.	Leukemia, acute myelogenous (JAK2)
2029.	Leukemia, acute myeloid (KIT)
2030.	Leukemia, acute myeloid (RUNX1)
2031.	Leukemia, acute myeloid form, susceptible due to TERT germline mutation (TERT)
2032.	Leukemia, acute myeloid, somatic (CEBPA)
2033.	Leukemia, juvenile myelomonocytic (PTPN11)
2034.	Leukemia, lymphoblastic and myeloid, EZH2 related (EZH2)
2035.	Leukemia, megakaryoblastic, with or without Down syndrome, somatic (GATA1)
2036.	Leukemia, myeloid acute form, due to CEBPA germline mutation (CEBPA)
2037.	Leukocyte adhesion deficiency (ITGB1)
2038.	Leukocyte adhesion deficiency (ITGB2)
2039.	Leukocyte adhesion deficiency type 3 (FERMT3)
2040.	Leukodystrophy demyelinating adult-onset, autosomal dominant (LMNB1)
2041.	Leukodystrophy hypomyelinating (GJC2)
2042.	Leukodystrophy hypomyelinating type 3 (AIMP1)
2043.	Leukodystrophy hypomyelinating type 4 (HSPD1)
2044.	Leukodystrophy hypomyelinating type 5 (FAM126A)
2045.	Leukodystrophy hypomyelinating type 6 (TUBB4A)
2046.	Leukodystrophy hypomyelinating type 7 (POLR3A)

2047.	Leukodystrophy hypomyelinating type 8 (POLR3B)
2048.	Leukodystrophy hypomyelinating type 9 (RARS)
2049.	Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (DARS2)
2050.	Leukoencephalopathy with dystonia and motor neuropathy (SCP2)
2051.	Leukoencephalopathy with vanishing white matter (EIF2B2)
2052.	Leukoencephalopathy with vanishing white matter (EIF2B1)
2053.	Leukoencephalopathy with vanishing white matter (EIF2B3)
2054.	Leukoencephalopathy with vanishing white matter (EIF2B4)
2055.	Leukoencephalopathy with vanishing white matter (EIF2B5)
2056.	Leukoencephalopathy, cystic without megalencephaly (RNASET2)
2057.	Leukoencephalopathy, diffuse hereditary, with spheroids (CSF1R)
2058.	Leukoencephalopathy, progressive, with ovarian failure (AARS2)
2059.	Lewy body dementia, susceptibility to (GBA)
2060.	Leydig cell hypoplasia type 1 (LHCGR)
2061.	Li-Fraumeni syndrome type 1 (TP53)
2062.	Li-Fraumeni syndrome type 2 (CHEK2)
2063.	Liddle syndrome (SCNN1B)
2064.	Liddle syndrome (SCNN1G)
2065.	LIG4 syndrome (LIG4)
2066.	Limb-girdle muscular dystrophy, autosomal dominant type 1A (MYOT)
2067.	Limb-girdle muscular dystrophy, autosomal dominant type 1B (LMNA)
2068.	Limb-girdle muscular dystrophy, autosomal dominant type 1C (CAV3)
2069.	Limb-girdle muscular dystrophy, autosomal dominant type 1E (DNAJB6)
2070.	Limb-girdle muscular dystrophy, autosomal recessice type 2F (SGCD)
2071.	Limb-girdle muscular dystrophy, autosomal recessive type 12C (POMK)
2072.	Limb-girdle muscular dystrophy, autosomal recessive type 2A (CAPN3)
2073.	Limb-girdle muscular dystrophy, autosomal recessive type 2B (DYSF)
2074.	Limb-girdle muscular dystrophy, autosomal recessive type 2C (SGCG)
2075.	Limb-girdle muscular dystrophy, autosomal recessive type 2D (SGCA)
2076.	Limb-girdle muscular dystrophy, autosomal recessive type 2E (SGCB)
2077.	Limb-Girdle Muscular Dystrophy, autosomal recessive type 2G (TCAP)
2078.	Limb-girdle muscular dystrophy, autosomal recessive type 2H (TRIM32)

2079.	Limb-girdle muscular dystrophy, autosomal recessive type 2I (FKRP)
2080.	Limb-girdle muscular dystrophy, autosomal recessive type 2J (NGS Sequencing: TTN)
2081.	Limb-girdle muscular dystrophy, autosomal recessive type 2K (POMT1)
2082.	Limb-girdle muscular dystrophy, autosomal recessive type 2L (ANO5)
2083.	Limb-girdle muscular dystrophy, autosomal recessive type 2M (FKTN)
2084.	Limb-girdle muscular dystrophy, autosomal recessive type 2N (POMT1)
2085.	Limb-girdle muscular dystrophy, autosomal recessive type 2S (TRAPPC11)
2086.	Limb-mammary syndrome (TP63)
2087.	Lipodystrophy generalized type 1 (AGPAT2)
2088.	Lipodystrophy generalized type 2 (BSCL2)
2089.	Lipodystrophy generalized type 4 (PTRF)
2090.	Lipodystrophy type 2, familial partial (LMNA)
2091.	Lipodystrophy, familial partial, type 3 (PPARG)
2092.	Lipoid congenital adrenal hyperplasia (STAR)
2093.	Lipoma, somatic (MEN1)
2094.	Lipoprotein glomerulopathy (APOE)
2095.	Lissencephaly type 1 (PAFAH1B1)
2096.	Lissencephaly type 2 (Norman-Roberts type) (NGS Sequencing: RELN)
2097.	Lissencephaly type 3 (TUBA1A)
2098.	Lissencephaly type 4 with microcephaly (NDE1)
2099.	Lissencephaly type 5 (LAMB1)
2100.	Lissencephaly, X-linked type 1 (DCX)
2101.	Lissencephaly, X-linked type 2 (ARX)
2102.	Lissencephaly/Subcortical laminal heteropia, X-linked (DCX)
2103.	Liver failure transient infantile (TRMU)
2104.	Loeys-Dietz syndrome type 1A (TGFBR1)
2105.	Loeys-Dietz syndrome type 1B (TGFBR2)
2106.	Loeys-Dietz syndrome type 1C (SMAD3)
2107.	Loeys-Dietz syndrome type 2A (TGFBR1)
2108.	Loeys-Dietz syndrome type 2B (TGFBR2)
2109.	Loeys-Dietz syndrome type 4 (TGFB2)
2110.	Long QT syndrome type 1 (KCNQ1)

2111.	Long QT syndrome type 10 (SCN4B)
2112.	Long QT syndrome type 11 (AKAP9)
2113.	Long QT syndrome type 12 (SNTA1)
2114.	Long QT syndrome type 13 (KCNJ5)
2115.	Long QT syndrome type 15 (CALM2)
2116.	Long QT syndrome type 2 (KCNH2)
2117.	Long QT syndrome type 3 (SCN5A)
2118.	Long QT syndrome type 4 (ANK2)
2119.	Long QT syndrome type 5 (KCNE1)
2120.	Long QT syndrome type 6 (KCNE2)
2121.	Long QT syndrome type 8 (CACNA1C)
2122.	Long QT syndrome type 9 (CAV3)
2123.	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (HADHA)
2124.	Lowe oculocerebrorenal syndrome (OCRL)
2125.	Lujan-Fryns syndrome (MED12)
2126.	Lung alpha-beta hydrolase deficiency type 1 (ABHD1)
2127.	Lung cancer, somatic (KRAS)
2128.	Lung cancer, SOX2 related, somatic (SOX2)
2129.	Lutheran inhibitor blood group (KLF1)
2130.	Lymphangioleiomyomatosis, somatic (TSC2)
2131.	Lymphedema, hereditary, type 1A (FLT4)
2132.	Lymphedema, hereditary, type IC (GJC2)
2133.	Lymphedema-distichiasis syndrome (FOXC2)
2134.	Lymphoma, B-cell type (BCL6)
2135.	Lymphoma, follicular, somatic (BCL10)
2136.	Lymphoma, MALT, somatic (BCL10)
2137.	Lymphoproliferative syndrome type 1 (ITK)
2138.	Lymphoproliferative syndrome type 2 (CD27)
2139.	Lymphoproliferative syndrome, autoimmune, type 5 (CTLA4)
2140.	Lymphoproliferative syndrome, X-linked type 1 (SH2D1A)
2141.	Lymphoproliferative syndrome, X-linked type 2 (XIAP)
2142.	Lynch syndrome-like tumors, MLH1 related, somatic (MLH1)

2143.	LYSINURIC PROTEIN INTOLERANCE (SLC7A7)
2144.	Lysosomal acid phosphatase deficiency (ACP2)
2145.	Macrocephaly, alopecia, cutis laxa, and scoliosis (RIN2)
2146.	Macroglobulinemia, Waldenstrom, somatic (MYD88)
2147.	Macular degeneration, age-related type 11 (CST3)
2148.	Macular degeneration, age-related type 3 (FBLN5)
2149.	Macular degeneration, age-related type 6 (RAX2)
2150.	Macular degeneration, age-related type 8, association with (ARMS2)
2151.	Macular degeneration, early-onset (NGS Sequencing: FBN2)
2152.	Macular dystrophy retinal type 2 (PROM1)
2153.	Macular dystrophy with central cone involvement (MFSD8)
2154.	Macular dystrophy, BEST2-related (BEST2)
2155.	Macular dystrophy, BEST3-related (BEST3)
2156.	Macular dystrophy, BEST4-related (BEST4)
2157.	Macular dystrophy, vitelliform (BEST1)
2158.	Macular dystrophy, vitelliform (PRPH2)
2159.	Mainzer Saldino syndrome (IFT140)
2160.	Majeed syndrome (LPIN2)
2161.	Major affective disorder (CUX2)
2162.	Major histocompatibility comples 1 deficiency (MR1)
2163.	Mal de Meleda (SLURP1)
2164.	Male germ cell tumor, somatic (BCL10)
2165.	Malignant hyperthermia type 5 (CACNA1S)
2166.	Malonyl-CoA decarboxylase deficiency (MLYCD)
2167.	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (POLD1)
2168.	Mandibuloacral dysplasia (LMNA)
2169.	Mandibuloacral dysplasia with type B lipodystrophy (ZMPSTE24)
2170.	Mandibulofacial dysostosis with microcephaly (EFTUD2)
2171.	Mannose-binding protein deficiency (MBL2)
2172.	Mannosidosis, beta A, lysosomal-like (MANBAL)
2173.	Mannosidosis-alpha (MAN2B1)
2174.	Mannosidosis-beta (MANBA)

2175.	Maple syrup urine disease type 1a (BCKDHA)
2176.	Maple syrup urine disease type 1b (BCKDHB)
2177.	Maple syrup urine disease type 2 (DBT)
2178.	Maple syrup urine disease type 3 (DLD)
2179.	Maple syrup urine disease, mild variant (PPM1K)
2180.	Marden-Walker syndrome (PIEZO2)
2181.	Marfan lipodystrophy syndrome (NGS Sequencing: FBN1)
2182.	Marfan syndrome (NGS Sequencing: FBN1)
2183.	Marfan syndrome, TGFBR1 related (TGFBR1)
2184.	Marfan syndrome, TGFBR2 related (TGFBR2)
2185.	Marshall syndrome (NGS Sequencing: COL11A1)
2186.	Martsolf syndrome (RAB3GAP2)
2187.	MASA syndrome (L1CAM)
2188.	MASS syndrome (NGS Sequencing: FBN1)
2189.	Maturity-onset diabetes of the young type 1 (HNF4A)
2190.	Maturity-onset diabetes of the young type 10 (INS)
2191.	Maturity-onset diabetes of the young type 11 (BLK)
2192.	Maturity-onset diabetes of the young type 2 (GCK)
2193.	Maturity-onset diabetes of the young type 3 (HNF1A)
2194.	Maturity-onset diabetes of the young type 4 (PDX1)
2195.	Maturity-onset diabetes of the young type 5 (HNF1B)
2196.	Maturity-onset diabetes of the young type 6 (NEUROD1)
2197.	Maturity-onset diabetes of the young type 7 (KLF11)
2198.	Maturity-onset diabetes of the young type 8 (CEL)
2199.	Maturity-onset diabetes of the young type 9 (PAX4)
2200.	Maturity-onset diabetes of the young, NKX2-2 related (NKX2-2)
2201.	Maturity-onset diabetes of the young, RFX6 related (RFX6)
2202.	Maturity-onset diabetes of the young, ZFP57 related (ZFP57)
2203.	McKusick-Kaufman syndrome (MKKS)
2204.	McLeod syndrome with or without chronic granulomatous disease (XK)
2205.	Meckel syndrome type 1 (MKS1)
2206.	Meckel syndrome type 10 (B9D2)

2207.	Meckel syndrome type 3 (TMEM67)
2208.	Meckel syndrome type 4 (CEP290)
2209.	Meckel syndrome type 8 (TCTN2)
2210.	Meckel syndrome type 9 (B9D1)
2211.	Meconium ileus (GUCY2C)
2212.	Mediterranean fever (MEFV)
2213.	MEDNIK syndrome (AP1S1)
2214.	Medullary cystic kidney disease type 2 (UMOD)
2215.	Medullary thyroid carcinoma, somatic (RET)
2216.	Medulloblastoma, desmoplastic, familial (SUFU)
2217.	Medulloblastoma, due to BRCA2 germline mutation (BRCA2)
2218.	Megalencephalic leukoencephalopathy with subcortical cysts (MLC1)
2219.	Megalencephalic leukoencephalopathy with subcortical cysts 2A (HEPACAM)
2220.	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome (PIK3R2)
2221.	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome type 2 (AKT3)
2222.	Megaloblastic anemia type 1 (AMN)
2223.	Megaloblastic anemia type 1, Finnish type (NGS Sequencing: CUBN)
2224.	Megalocornea, X-linked (CHRDL1)
2225.	Meier-Gorlin syndrome 4 (CDT1)
2226.	Meier-Gorlin syndrome type 1 (ORC1)
2227.	Melanocytic nevus syndrome, congenital, somatic (NRAS)
2228.	Melanoma and neural system tumor syndrome, familial (CDKN2A)
2229.	Melanoma, cutaneous malignant (MC1R)
2230.	Melanoma, cutaneous malignant (MITF)
2231.	Melanoma, cutaneous malignant, familial (CDKN2A)
2232.	Melanoma, cutaneous malignant, familial type 10, susceptibility to (POT1)
2233.	Melanoma, cutaneous malignant, familial type 6, susceptibility to (XRCC3)
2234.	Melanoma, cutaneous malignant, familial, CDK4 related (CDK4)
2235.	Melanoma, malignant, somatic (BRAF)
2236.	Melanoma, malignant, somatic (STK11)
2237.	MELAS syndrome (NGS Sequencing: MT-TF)
2238.	MELAS syndrome (NGS Sequencing: MT-TC)

2239.	MELAS syndrome, MT-TL1 related (NGS Sequencing: MT-TL1)
2240.	Melnick-Needles syndrome (FLNA)
2241.	Meningioma, familial, PDGFB related (PDGFB)
2242.	Meningioma, familial, susceptibility to (SUFU)
2243.	Meningioma, familial, susceptibility to (SMARCE1)
2244.	Meningioma, MN1 deficiency related (MN1)
2245.	Meningioma, NF2-related, somatic (NF2)
2246.	Menkes disease (ATP7A)
2247.	Mental retardation and distinctive facial features with or without cardiac defects (MED13L)
2248.	Mental retardation and microcephaly with pontine and cerebellar hypoplasia (CASK)
2249.	Mental retardation non-syndromic (KLF8)
2250.	Mental retardation non-syndromic (NXF5)
2251.	Mental retardation non-syndromic (ZCCHC12)
2252.	Mental retardation non-syndromic (ELK1)
2253.	Mental retardation with Cerebellar ataxia and dysequilibrium syndrome type 2 (WDR81)
2254.	Mental retardation with hypotonic facies syndrome, X-linked (ATRX)
2255.	Mental retardation with language impairment and autistic features (FOXP1)
2256.	Mental retardation X-linked, SMARCA1 related (SMARCA1)
2257.	Mental retardation X-linked, syndromic, Claes-Jensen type (KDM5C)
2258.	Mental retardation X-linked, syndromic, Lubs type (MECP2)
2259.	Mental retardation, autosomal dominant type 1 (MBD5)
2260.	Mental retardation, autosomal dominant type 12 (ARID1B)
2261.	Mental retardation, autosomal dominant type 13 (NGS Sequencing: DYNC1H1)
2262.	Mental retardation, autosomal dominant type 13 (TRAPPC9)
2263.	Mental retardation, autosomal dominant type 14 (ARID1A)
2264.	Mental retardation, autosomal dominant type 15 (SMARCB1)
2265.	Mental retardation, autosomal dominant type 16 (SMARCA4)
2266.	Mental retardation, autosomal dominant type 17 (PACS1)
2267.	Mental retardation, autosomal dominant type 18 (GATAD2B)
2268.	Mental retardation, autosomal dominant type 19 (CTNNB1)
2269.	Mental retardation, autosomal dominant type 2 (DOCK8)
2270.	Mental retardation, autosomal dominant type 20 (MEF2C)

2271.	Mental retardation, autosomal dominant type 23 (SETD5)
2272.	Mental retardation, autosomal dominant type 24 (DEAF1)
2273.	Mental retardation, autosomal dominant type 25 (AHDC1)
2274.	Mental retardation, autosomal dominant type 27 (SOX11)
2275.	Mental retardation, autosomal dominant type 28 (ADNP)
2276.	Mental retardation, autosomal dominant type 31 (PURA)
2277.	Mental retardation, autosomal dominant type 32 (KAT6A)
2278.	Mental retardation, autosomal dominant type 37 (POGZ)
2279.	Mental retardation, autosomal dominant type 38 (EEF1A2)
2280.	Mental retardation, autosomal dominant type 5 (SYNGAP1)
2281.	Mental retardation, autosomal dominant type 6 (GRIN2B)
2282.	Mental retardation, autosomal dominant type 7 (DYRK1A)
2283.	Mental retardation, autosomal dominant type 8 (GRIN1)
2284.	Mental retardation, autosomal dominant type 9 (KIF1A)
2285.	Mental retardation, autosomal recessive type 12 (ST3GAL3)
2286.	Mental retardation, autosomal recessive type 14 (TECR)
2287.	Mental retardation, autosomal recessive type 15 (MAN1B1)
2288.	Mental retardation, autosomal recessive type 18 (MED23)
2289.	Mental retardation, autosomal recessive type 2 (CRBN)
2290.	Mental retardation, autosomal recessive type 27 (LINS1)
2291.	Mental retardation, autosomal recessive type 3 (CC2D1A)
2292.	Mental retardation, autosomal recessive type 36 (ADAT3)
2293.	Mental retardation, autosomal recessive type 37 (NGS Sequencing: ANK3)
2294.	Mental retardation, autosomal recessive type 38 (NGS Sequencing: HERC2)
2295.	Mental retardation, autosomal recessive type 39 (TTI2)
2296.	Mental retardation, autosomal recessive type 41 (KPTN)
2297.	Mental retardation, autosomal recessive type 42 (PGAP1)
2298.	Mental retardation, autosomal recessive type 46 (NDST1)
2299.	Mental retardation, autosomal recessive type 49 (GPT2)
2300.	Mental retardation, autosomal recessive type 5 (NSUN2)
2301.	Mental retardation, autosomal recessive type 7 (TUSC3)
2302.	Mental retardation, truncal obesity, retinal dystrophy, and micropenis (INPP5E)

2303.	Mental retardation, X-linked (RAB40AL)
2304.	Mental retardation, X-linked syndromic, Christianson type (SLC9A6)
2305.	Mental retardation, X-linked syndromic, Nascimento-type (UBE2A)
2306.	Mental retardation, X-linked syndromic, Raymond type (ZDHHC9)
2307.	Mental retardation, X-linked syndromic, Turner type (NGS Sequencing: HUWE1)
2308.	Mental retardation, X-linked type 1 (IQSEC2)
2309.	Mental retardation, X-linked type 101 (MID2)
2310.	Mental retardation, X-linked type 102 (DDX3X)
2311.	Mental retardation, X-Linked type 13 (MECP2)
2312.	Mental retardation, X-linked type 14 (UPF3B)
2313.	Mental retardation, X-linked type 15 (CUL4B)
2314.	Mental retardation, X-linked type 16 (FGD1)
2315.	Mental retardation, X-linked type 17 (HSD17B10)
2316.	Mental retardation, X-linked type 19 (RPS6KA3)
2317.	Mental retardation, X-linked type 21 (IL1RAPL1)
2318.	Mental retardation, X-linked type 29 (ARX)
2319.	Mental retardation, X-linked type 3 (HCFC1)
2320.	Mental retardation, X-linked type 30 (PAK3)
2321.	Mental retardation, X-linked type 32 (CLIC2)
2322.	Mental retardation, X-linked type 41 (GDI1)
2323.	Mental retardation, X-linked type 44 (FTSJ1)
2324.	Mental retardation, X-linked type 45 (ZNF81)
2325.	Mental retardation, X-linked type 46 (ARHGEF6)
2326.	Mental retardation, X-linked type 58 (TSPAN7)
2327.	Mental retardation, X-linked type 59 (AP1S2)
2328.	Mental retardation, X-linked type 63 (ACSL4)
2329.	Mental retardation, X-linked type 72 (RAB39B)
2330.	Mental retardation, X-linked type 88, AGTR2 related (AGTR2)
2331.	Mental retardation, X-linked type 89 (ZNF41)
2332.	Mental retardation, X-linked type 90 (DLG3)
2333.	Mental retardation, X-linked type 91 (ZDHHC15)
2334.	Mental retardation, X-linked type 92 (ZNF674)

2335.	Mental retardation, X-linked type 93 (BRWD3)
2336.	Mental retardation, X-linked type 94 (GRIA3)
2337.	Mental retardation, X-linked type 95 (MAGT1)
2338.	Mental retardation, X-linked type 96 (SYP)
2339.	Mental retardation, X-linked type 97 (ZNF711)
2340.	Mental retardation, X-linked type 99 (USP9X)
2341.	Mental retardation, X-linked with epilepsy (ATP6AP2)
2342.	Mental retardation, X-linked, associated with fragile site FRAXE (AFF2)
2343.	Mental retardation, x-linked, EFHC2 related (EFHC2)
2344.	Mental retardation, X-linked, nonsyndromic (KIAA2022)
2345.	Mental retardation, X-linked, Siderius type (PHF8)
2346.	Mental retardation, X-linked, Snyder-Robinson type (SMS)
2347.	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance (OPHN1)
2348.	Mental retardation, X-linked, with isolated growth hormone deficiency (SOX3)
2349.	MERRF syndrome, MT-TK related (NGS Sequencing: MT-TK)
2350.	MERRF syndrome, MT-TP related (NGS Sequencing: MT-TP)
2351.	MERRF/MELAS overlap syndrome, MT-TS1 related (NGS Sequencing: MT-TS1)
2352.	MERRF/MELAS overlap syndrome, MT-TS2 related (NGS Sequencing: MT-TS2)
2353.	Mesothelioma, somatic (BCL10)
2354.	Mesothelioma, somatic (WT1)
2355.	Metachromatic Leukodystrophy (ARSA)
2356.	Metachromatic leukodystrophy due to Saposin B deficiency (PSAP)
2357.	Metaphyseal anadysplasia type 1 (MMP13)
2358.	Metaphyseal anadysplasia type 2 (MMP9)
2359.	Metaphyseal chondrodysplasia, Schmid type (COL10A1)
2360.	Metaphyseal chondromatosis with increased urinary excretion of D-2-hydroxyglutarate (IDH1)
2361.	Metaphyseal dysplasia without hypotrichosis (RMRP)
2362.	Methemoglobinemia type 1 (CYB5R3)
2363.	Methionine adenosyltransferase deficiency, autosomal recessive (MAT1A)
2364.	Methylacetoacetic aciduria (ACAT1)
2365.	Methylcobalamin deficiency CblG type (MTR)
2366.	Methylmalonate semialdehyde dehydrogenase deficiency (ALDH6A1)

2367.	Methylmalonic aciduria CblA type (MMAA)
2368.	Methylmalonic aciduria CblB type (MMAB)
2369.	Methylmalonic aciduria CblC type (MMACHC)
2370.	Methylmalonic aciduria CblD type (MMADHC)
2371.	Methylmalonic aciduria CblF type (LMBRD1)
2372.	Methylmalonic aciduria CblJ type (ABCD4)
2373.	Methylmalonic aciduria CblR type (CD320)
2374.	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency (MUT)
2375.	Methylmalonyl-CoA epimerase deficiency (MCEE)
2376.	Mevalonic aciduria (MVK)
2377.	Microcephalic osteodysplastic primordial dwarfism type 1 (RNU4ATAC)
2378.	Microcephalic osteodysplastic primordial dwarfism type 2 (PCNT)
2379.	Microcephaly and chorioretinopathy with or without mental retardation (TUBGCP6)
2380.	Microcephaly with cortical malformations, autosomal recessive type 2 (WDR62)
2381.	Microcephaly with epilepsy and diabetes syndrome (IER3IP1)
2382.	Microcephaly with or without chorioretinopathy, Lymphedema, or Mental retardation, MCLMR (KIF11)
2383.	Microcephaly with symplified gyral pattern and insulin-dependant diabetes (GFM2)
2384.	Microcephaly, Amish type (SLC25A19)
2385.	Microcephaly, AP4M1 related (AP4M1)
2386.	Microcephaly, autosomal recessive type 1 (MCPH1)
2387.	Microcephaly, autosomal recessive type 11 (PHC1)
2388.	Microcephaly, autosomal recessive type 12 (CDK6)
2389.	Microcephaly, autosomal recessive type 13 (CENPE)
2390.	Microcephaly, autosomal recessive type 3 (CDK5RAP2)
2391.	Microcephaly, autosomal recessive type 4 (CASC5)
2392.	Microcephaly, autosomal recessive type 5 (ASPM)
2393.	Microcephaly, autosomal recessive type 6 (CENPJ)
2394.	Microcephaly, autosomal recessive type 7 (STIL)
2395.	Microcephaly, autosomal recessive type 8 (CEP135)
2396.	Microcephaly, autosomal recessive type 9 (CEP152)
2397.	Microcephaly, CEP63 related (CEP63)
2398.	Microcephaly, MRE11A related (MRE11A)

2399.	Microcephaly, MSMO1 related (MSMO1)
2400.	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy (QARS)
2401.	Microcephaly, short stature, and polymicrogyria with seizures (RTTN)
2402.	Microcephaly, TUBB2B related (TUBB2B)
2403.	Microcephaly-capillary malformation syndrome (STAMBP)
2404.	Microphthalmia syndromic type 2 (BCOR)
2405.	Microphthalmia syndromic type 3 (SOX2)
2406.	Microphthalmia syndromic type 5 (OTX2)
2407.	Microphthalmia syndromic type 6 (BMP4)
2408.	Microphthalmia syndromic type 6 (SIX6)
2409.	Microphthalmia syndromic type 7 (HCCS)
2410.	Microphthalmia syndromic type 8 (ALDH1A3)
2411.	Microphthalmia syndromic type 9 (STRA6)
2412.	Microphthalmia, isolated type 2 (VSX2)
2413.	Microphthalmia, isolated type 3 (RAX)
2414.	Microphthalmia, isolated type 4 (GDF6)
2415.	Microphthalmia, isolated type 5 (MFRP)
2416.	Microphthalmia, isolated type 6 (PRSS56)
2417.	Microphthalmia, isolated type 9 (GDF3)
2418.	Microphthalmia, isolated with coloboma type 3 (VSX2)
2419.	Microphthalmia, isolated with coloboma type 6, digenic (GDF6)
2420.	Microphthalmia, isolated with coloboma type 6, digenic (GDF3)
2421.	Microphthalmia, isolated with coloboma type 9 (TENM3)
2422.	Microphthalmia, syndromic type 1 (NAA10)
2423.	Microphthalmia, syndromic type 11 (VAX1)
2424.	Microphthalmia, VAX2 related (VAX2)
2425.	Microspherophakia and/or megalocornea (LTBP2)
2426.	Microvascular complications of diabetes type 1 (VEGFA)
2427.	Microvascular complications of diabetes type 6, susceptibility to (SOD2)
2428.	Miller Dieker lissencephaly syndrome (YWHAE)
2429.	Minicore myopathy with external ophthalmoplegia (NGS Sequencing: RYR1)
2430.	Mirror movements type 1 (DCC)

2431.	Mirror movements type 2 (RAD51)
2432.	Mirror movements type 3 (DNAL4)
2433.	Mismatch repair cancer syndrome (MSH6)
2434.	Mismatch repair cancer syndrome (PMS2)
2435.	Mismatch repair cancer syndrome (MLH1)
2436.	Mismatch repair cancer syndrome (MSH2)
2437.	Mitchell-Riley syndrome (RFX6)
2438.	Mitochondrial complex I deficiency (FOXRED1)
2439.	Mitochondrial complex I deficiency (NDUFB3)
2440.	Mitochondrial complex I deficiency (NDUFS2)
2441.	Mitochondrial complex I deficiency (NDUFS1)
2442.	Mitochondrial complex I deficiency (NDUFAF4)
2443.	Mitochondrial complex I deficiency (NGS Sequencing: MT-ND4)
2444.	Mitochondrial complex I deficiency (NGS Sequencing: MT-ND3)
2445.	Mitochondrial complex I deficiency (NGS Sequencing: MT-ND2)
2446.	Mitochondrial complex I deficiency (NGS Sequencing: MT-ND6)
2447.	Mitochondrial complex I deficiency (NGS Sequencing: MT-ND1)
2448.	Mitochondrial complex I deficiency (NGS Sequencing: MT-ND5)
2449.	Mitochondrial complex I deficiency (NGS Sequencing: MT-ND4L)
2450.	Mitochondrial complex I deficiency (NDUFA1)
2451.	Mitochondrial complex I deficiency (NDUFAF3)
2452.	Mitochondrial complex I deficiency (NDUFAF5)
2453.	Mitochondrial complex I deficiency (NDUFAF1)
2454.	Mitochondrial complex I deficiency (NDUFS6)
2455.	Mitochondrial complex I deficiency (NDUFS4)
2456.	Mitochondrial complex I deficiency (NDUFV2)
2457.	Mitochondrial complex I deficiency (NDUFV1)
2458.	Mitochondrial complex I deficiency (NDUFA11)
2459.	Mitochondrial complex I deficiency, MT-TN related (NGS Sequencing: MT-TN)
2460.	Mitochondrial complex II deficiency (SDHAF1)
2461.	Mitochondrial complex II deficiency (SDHD)
2462.	Mitochondrial complex III deficiency (UQCRC2)

2463.	Mitochondrial complex III deficiency (UQCRQ)
2464.	Mitochondrial complex III deficiency (UQCRB)
2465.	Mitochondrial complex III deficiency (BCS1L)
2466.	Mitochondrial complex III deficiency, nuclear type 2 (TTC19)
2467.	Mitochondrial complex III deficiency, nuclear type 7 (UQCC2)
2468.	Mitochondrial complex IV deficiency (COX6B1)
2469.	Mitochondrial complex IV deficiency (FASTKD2)
2470.	Mitochondrial complex IV deficiency (PET100)
2471.	Mitochondrial complex IV deficiency (APOPT1)
2472.	Mitochondrial complex IV deficiency (NGS Sequencing: MT-CO3)
2473.	Mitochondrial complex V (ATP synthase) deficiency (NGS Sequencing: MT-ATP6)
2474.	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1 (ATPAF2)
2475.	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2 (TMEM70)
2476.	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3 (ATP5E)
2477.	Mitochondrial complex V deficiency, nuclear type 4 (ATP5A1)
2478.	Mitochondrial Disorders, AKAP1 related (AKAP1)
2479.	Mitochondrial DNA depletion syndrome (SUCLA2)
2480.	Mitochondrial DNA depletion syndrome (TK2)
2481.	Mitochondrial DNA depletion syndrome (DGUOK)
2482.	Mitochondrial DNA depletion syndrome 8B, MNGIE type (RRM2B)
2483.	Mitochondrial DNA depletion syndrome type 11 (MGME1)
2484.	Mitochondrial DNA depletion syndrome type 13 (FBXL4)
2485.	Mitochondrial DNA depletion syndrome type 4A (POLG)
2486.	Mitochondrial DNA depletion syndrome type 4B (POLG)
2487.	Mitochondrial DNA depletion syndrome type 6 (MPV17)
2488.	Mitochondrial DNA depletion syndrome type 7 (TWNK)
2489.	Mitochondrial DNA depletion syndrome, encephalomyopathic type with methylmalonic aciduria (SUCLG1)
2490.	Mitochondrial encephalomyopathy (MFF)
2491.	Mitochondrial encephalomyopathy (NGS Sequencing: MT-CYB)
2492.	Mitochondrial modifier of deafness (TRMU)
2493.	Mitochondrial myopathy and sideroblastic anemia type 1 (PUS1)
2494.	Mitochondrial myopathy, infantile, transient, MT-TE related (NGS Sequencing: MT-TE)

2495.	Mitochondrial myopathy, isolated (NGS Sequencing: MT-TD)
2496.	Mitochondrial myopathy, MT-TA related (NGS Sequencing: MT-TA)
2497.	Mitochondrial myopathy, MT-TM related (NGS Sequencing: MT-TM)
2498.	Mitochondrial neurogastrointestinal encephalopathy syndrome without leukoencephalopathy (TYMP)
2499.	Mitochondrial neurogastrointestinal encephalopathy syndrome without leukoencephalopathy (POLG)
2500.	Mitochondrial pyruvate carrier deficiency (MPC1)
2501.	Mitochondrial respiratory chain complex II deficiency (SDHA)
2502.	Mitochondrial respiratory chain disease, TIMM21 related (TIMM21)
2503.	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency (ECHS1)
2504.	Miyoshi muscular dystrophy type 3 (ANO5)
2505.	Miyoshi myopathy (DYSF)
2506.	MMR genes methylation analysis (MMR genes)
2507.	Molybdenum cofactor deficiency type A (MOCS1)
2508.	Molybdenum cofactor deficiency type B (MOCS2)
2509.	Molybdenum cofactor deficiency type C (GPHN)
2510.	Monocarboxylate transporter 1 deficiency (SLC16A1)
2511.	Mosaic variegated aneuploidy syndrome type 2 (CEP57)
2512.	Moyamoya disease type 2, susceptibility to (NGS Sequencing: RNF213)
2513.	Moyamoya disease type 5 (ACTA2)
2514.	Moyamoya type 6 with achalasia (GUCY1A3)
2515.	MPL, selective sequencing of exon 10 (MPL)
2516.	Muckle-wells syndrome (NLRP3)
2517.	Mucolipidosis type 2 alpha/beta (GNPTAB)
2518.	Mucolipidosis type 3 (GNPTAB)
2519.	Mucolipidosis type 3 gamma (GNPTG)
2520.	Mucolipidosis type 4 (MCOLN1)
2521.	Mucopolysaccharidosis type 2 (IDS)
2522.	Mucopolysaccharidosis type 3A (SGSH)
2523.	Mucopolysaccharidosis type 3B (NAGLU)
2524.	Mucopolysaccharidosis type 3C (HGSNAT)
2525.	Mucopolysaccharidosis type 3D (GNS)
2526.	Mucopolysaccharidosis type 4A (GALNS)

2527.	Mucopolysaccharidosis type 4B (GLB1)
2528.	Mucopolysaccharidosis type 6 (ARSB)
2529.	Mucopolysaccharidosis type 7 (GUSB)
2530.	Mucopolysaccharidosis type 9 (HYAL1)
2531.	Mucopolysaccharidosis type IH (IDUA)
2532.	Muir-Torre syndrome (MLH1)
2533.	Muir-Torre syndrome (MSH2)
2534.	Mulibrey nanism (TRIM37)
2535.	Multicentric carpotarsal osteolysis syndrome (MAFB)
2536.	Multicentric osteolysis, nodulosis, and arthropathy (MMP2)
2537.	Multiple congenital anomalies-hypotonia-seizures syndrome type 1 (PIGN)
2538.	Multiple congenital anomalies-hypotonia-seizures syndrome type 3 (PIGT)
2539.	Multiple endocrine neoplasia type 1 (MEN1)
2540.	Multiple endocrine neoplasia type 1, CDKN2B related (CDKN2B)
2541.	Multiple endocrine neoplasia type 2A (RET)
2542.	Multiple endocrine neoplasia type 2B (RET)
2543.	Multiple endocrine neoplasia type 4 (CDKN1B)
2544.	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects (B3GAT3)
2545.	Multiple mitochondrial dysfunctions syndrome type 1 (NFU1)
2546.	Multiple mitochondrial dysfunctions syndrome type 2 (BOLA3)
2547.	Multiple mitochondrial dysfunctions syndrome type 3 (IBA57)
2548.	Multiple mitochondrial dysfunctions syndrome type 4 (ISCA2)
2549.	Multiple pterygium syndrome lethal type (CHRNA1)
2550.	Multiple pterygium syndrome lethal type (CHRND)
2551.	Multisystemic smooth muscle dysfunction syndrome (ACTA2)
2552.	Muscle glycogenosis (PHKA1)
2553.	Muscle hypertrophy (MSTN)
2554.	Muscle-eye-brain disease, POMK related (POMK)
2555.	Muscular dystrophy type 1A (LAMA2)
2556.	Muscular dystrophy type 1C (FKRP)
2557.	Muscular dystrophy type 1D (LARGE)
2558.	Muscular dystrophy, Becker type (NGS Sequencing: DMD)

2559.	Muscular dystrophy, congenital, LMNA related (LMNA)
2560.	Muscular dystrophy, congenital, megaconial type (CHKB)
2561.	Muscular dystrophy, Duchenne type (NGS Sequencing: DMD)
2562.	Muscular dystrophy, limb-girdle type 2A (CAPN3)
2563.	Muscular dystrophy, limb-girdle, type 2Q (NGS Sequencing: PLEC)
2564.	Muscular dystrophy, oculopharyngeal (PABPN1)
2565.	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A10 (TMEM5)
2566.	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A13 (B4GAT1)
2567.	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A2 (POMT2)
2568.	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A3 (POMGNT1)
2569.	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A8 (POMGNT2)
2570.	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B1 (POMT1)
2571.	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B2 (POMT2)
2572.	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B3 (POMGNT1)
2573.	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C2 (POMT2)
2574.	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C3 (POMGNT1)
2575.	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C9 (DAG1)
2576.	Muscular-skeletal disorder, CAPN1 related (CAPN1)
2577.	Myasthenia congenital with tubular aggregates 1 (GFPT1)
2578.	Myasthenic syndrome associated with acetylcholine receptor deficiency (MUSK)
2579.	Myasthenic syndrome due to mutation in SCN4A (SCN4A)
2580.	Myasthenic syndrome fast channel congenital (CHRNA1)
2581.	Myasthenic syndrome slow-channel congenital (CHRNA1)
2582.	Myasthenic syndrome type 10, congenital (DOK7)
2583.	Myasthenic syndrome type 11, congenital, associated with acetylcholine receptor deficiency (RAPSN)
2584.	Myasthenic syndrome, congenital (AGRN)
2585.	Myasthenic syndrome, congenital (CHRNB1)
2586.	Myasthenic syndrome, congenital (CHAT)
2587.	Myasthenic syndrome, congenital (CHRNE)
2588.	Myasthenic syndrome, congenital, type 3C, associated with acetylcholine receptor deficiency (CHRND)
2589.	Myasthenic syndrome, fast-channel, congenital, type 3B (CHRND)
2590.	Myasthenic syndrome, slow channel, congenital, type 3A (CHRND)

2591.	Mycobacterial infection, atypical, familial disseminated (IFNGR1)
2592.	Myelodysplastic syndrome, somatic (TET2)
2593.	Myelofibrosis, somatic (JAK2)
2594.	Myeloproliferative disorder, chronic, with eosinophilia (PDGFRB)
2595.	Myhre syndrome (SMAD4)
2596.	Myoclonic dystonia, DRD2 related (DRD2)
2597.	Myoclonic epilepsy of Lafora (EPM2A)
2598.	Myoclonic epilepsy of Lafora (NHLRC1)
2599.	Myoclonus, familial cortical (NOL3)
2600.	Myoglobinuria acute recurrent (LPIN1)
2601.	Myopathy due to Integrin 7A deficiency (ITGA7)
2602.	Myopathy due to myoadenylate deaminase deficiency (AMPD1)
2603.	Myopathy with fiber-type disproportion (SELENON)
2604.	Myopathy with fiber-type disproportion type 1 (ACTA1)
2605.	Myopathy with lactic acidosis hereditary (ISCU)
2606.	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset (MEGF10)
2607.	Myopathy, centronuclear (BIN1)
2608.	Myopathy, centronuclear (DNM2)
2609.	Myopathy, COL6A6 related (COL6A6)
2610.	Myopathy, desmin related, associated with mutation in the CRYAB gene (CRYAB)
2611.	Myopathy, distal type 1 (MYH7)
2612.	Myopathy, distal type 4 (FLNC)
2613.	Myopathy, distal with anterior tibial onset (DYSF)
2614.	Myopathy, distal, Tateyama type (CAV3)
2615.	Myopathy, early-onset with fatal cardiomyopathy (NGS Sequencing: TTN)
2616.	Myopathy, lactic acidosis, and sideroblastic anemia type 2 (YARS2)
2617.	Myopathy, limb girdle with bone fragility (MTAP)
2618.	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay (GFER)
2619.	Myopathy, MT-TQ related (NGS Sequencing: MT-TQ)
2620.	Myopathy, myofibrillar type 6 (BAG3)
2621.	Myopathy, myofibrillar, Desmin related (DES)
2622.	Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related (CRYAB)

2623.	Myopathy, myofibrillar, ZASP related (LDB3)
2624.	Myopathy, scapulohumeroperoneal (ACTA1)
2625.	Myopathy, tubular aggregate, type 1 (STIM1)
2626.	Myopathy, tubular aggregate, type 2 (ORAI1)
2627.	Myosclerosis, autosomal recessive (COL6A2)
2628.	Myosin storage myopathy (MYH7)
2629.	Myotilinopathy (MYOT)
2630.	Myotonia congenita (CLCN1)
2631.	Myotonic dystrophy type 1 (DMPK)
2632.	Myotonic dystrophy type 2 (CNBP)
2633.	Myotubular myopathy X-linked (MTM1)
2634.	Myxoma, intracardiac (PRKAR1A)
2635.	N-acetylglutamate synthase deficiency (NAGS)
2636.	Naegeli-Franceschetti-Jadassohn syndrome (KRT14)
2637.	Nail-Patella syndrome (LMX1B)
2638.	Nance-Horan syndrome (NHS)
2639.	Nanophthalmia type 2 (MFRP)
2640.	Narcolepsy (HCRT)
2641.	Nasopharyngeal carcinoma, somatic (TP53)
2642.	Native American myopathy (STAC3)
2643.	Nemaline myopathy type 1 (TPM3)
2644.	Nemaline myopathy type 2, autosomal recessive (NGS Sequencing: NEB)
2645.	Nemaline myopathy type 3 (ACTA1)
2646.	Nemaline myopathy type 4 (TPM2)
2647.	Nemaline myopathy type 5 (TNNT1)
2648.	Nemaline myopathy type 6 (KBTBD13)
2649.	Nemaline myopathy type 7 (CFL2)
2650.	Neonatal death due Leigh syndrome, MT-TV related (NGS Sequencing: MT-TV)
2651.	Nephrogenic syndrome of inapproriate antidiuresis (AVPR2)
2652.	Nephrolithiasis type 1 (CLCN5)
2653.	Nephrolithiasis/osteoporosis, hypophosphatemic, type 1 (SLC34A1)
2654.	Nephrolithiasis/osteoporosis, hypophosphatemic, type 2 (SLC9A3R1)

2655.	Nephronophthisis type 1 (NPHP1)
2656.	Nephronophthisis type 12 (TTC21B)
2657.	Nephronophthisis type 13 (WDR19)
2658.	Nephronophthisis type 14 (ZNF423)
2659.	Nephronophthisis type 15 (CEP164)
2660.	Nephronophthisis type 16 (ANKS6)
2661.	Nephronophthisis type 19 (DCDC2)
2662.	Nephronophthisis type 2 (INVS)
2663.	Nephronophthisis type 3 (NPHP3)
2664.	Nephronophthisis type 4 (NPHP4)
2665.	Nephronophthisis type 7 (GLIS2)
2666.	Nephronophthisis type 9 (NEK8)
2667.	Nephronophthisis-like nephropathy type 1 (XPNPEP3)
2668.	Nephrosis, Finnish type (NPHS1)
2669.	Nephrotic syndrome (NPHS2)
2670.	Nephrotic syndrome type 2 (NPHS1)
2671.	Nephrotic syndrome type 3 (PLCE1)
2672.	Nephrotic syndrome type 5 (LAMB2)
2673.	Nephrotic syndrome type 7 (DGKE)
2674.	Nephrotic syndrome type 8 (ARHGDIA)
2675.	Nephrotic syndrome type 9 (ADCK4)
2676.	Netherton syndrome (SPINK5)
2677.	Neuraminidase deficiency (NEU1)
2678.	Neuroaxonal neurodegeneration, infantile, with facial dysmophism (NALCN)
2679.	Neuroblastoma type 3, susceptibility to, familial (ALK)
2680.	Neurocutaneous melanosis, somatic (NRAS)
2681.	Neurodegeneration due to cerebral folate transport deficiency (FOLR1)
2682.	Neurodegeneration with brain iron accululation type 5 (WDR45)
2683.	Neurodegeneration with brain iron accumulation type 4 (C19orf12)
2684.	Neurodegeneration with brain iron accumulation type 6 (COASY)
2685.	Neurodegeneration with brain iron accumulation, GTPBP2 related (GTPBP2)
2686.	Neurodevelopmental disorder, ADAM22 related (ADAM22)

2687.	Neurodevelopmental disorder, APC2-related (APC2)
2688.	Neurodevelopmental disorder, CNTNAP4 related (CNTNAP4)
2689.	Neurodevelopmental disorder, CROCC related (CROCC)
2690.	Neurodevelopmental disorder, FRMPD4 related (FRMPD4)
2691.	Neurodevelopmental disorder, KCTD3 related (KCTD3)
2692.	Neurodevelopmental disorder, MACF1 related (NGS Sequencing: MACF1)
2693.	Neurodevelopmental disorder, MTOR related (MTOR)
2694.	Neurodevelopmental disorder, NCAM1 related (NCAM1)
2695.	Neurodevelopmental disorder, NGEF related (NGEF)
2696.	Neurodevelopmental disorder, PIGQ related (PIGQ)
2697.	Neurodevelopmental disorder, TUBB related (TUBB)
2698.	Neurodevelopmental disorder, ZNF311 related (ZNF311)
2699.	Neurodevelopmental malformation and microcephaly (TUBG1)
2700.	Neurodevelopmental malformation and microcephaly (KIF2A)
2701.	Neurodevelopmental malformation and microcephaly (KIF5C)
2702.	Neurofibromatosis type 1 (NGS Sequencing: NF1)
2703.	Neurofibromatosis type 1 -like syndrome (SPRED1)
2704.	Neurofibromatosis type 2 (NF2)
2705.	Neurogenic scapuloperoneal syndrome, Kaeser type (DES)
2706.	Neuromyotonia and axonal neuropathy, autosomal recessive (HINT1)
2707.	Neuronal migration disorder (NGS Sequencing: SPTBN5)
2708.	Neuronal migration disorder (EOMES)
2709.	Neuronal migration disorder (SRGAP2)
2710.	Neuronal migration disorder (CTNNA2)
2711.	Neuronopathy distal hereditary motor type 2A (HSPB8)
2712.	Neuronopathy distal hereditary motor type 2B (HSPB1)
2713.	Neuronopathy distal hereditary motor type 5 (GARS)
2714.	Neuronopathy distal hereditary motor type 6 (IGHMBP2)
2715.	Neuronopathy distal hereditary motor type 7B (DCTN1)
2716.	Neuropathy sensor type 1E (DNMT1)
2717.	Neuropathy with liability to pressure palsies [HNPP] (PMP22)
2718.	Neuropathy with sensory ataxic, dysarthria, and ophthalmoparesis (POLG)

2719.	Neuropathy, distal hereditary motor, type 5A (BSCL2)
2720.	Neuropathy, hereditary sensory and autonomic type 2 (SCN9A)
2721.	Neuropathy, hereditary sensory and autonomic type 6 (NGS Sequencing: DST)
2722.	Neuropathy, hereditary sensory, type ID (ATL1)
2723.	Neuropathy, hereditary sensory, with spastic paraplegia (CCT5)
2724.	Neutral lipid storage disease with myopathy (PNPLA2)
2725.	Neutropenia, nonimmune chronic idiopathic, of adults (GFI1)
2726.	Neutropenia, severe congenital type 1 (ELANE)
2727.	Neutropenia, severe congenital type 2, autosomal dominant (GFI1)
2728.	Neutropenia, severe congenital type 3 (HAX1)
2729.	Neutropenia, severe congenital type 4, autosomal recessive (G6PC3)
2730.	Neutropenia, severe congenital type 5, autosomal recessive (VPS45)
2731.	Neutropenia, severe congenital type 6, autosomal recessive (JAGN1)
2732.	Neutrophil immunodeficiency syndrome (RAC2)
2733.	Neutrophilia, hereditary (CSF3R)
2734.	Nevus sebaceous, HRAS related, somatic (HRAS)
2735.	Nevus, epidermal, somatic (FGFR3)
2736.	Nicolaides Baraitser syndrome (SMARCA2)
2737.	Niemann-Pick disease type A/B (SMPD1)
2738.	Niemann-Pick disease type C1 (NPC1)
2739.	Niemann-Pick disease type C2 (NPC2)
2740.	Night blindness type 1, congenital stationary, autosomal dominant (RHO)
2741.	Night blindness, congenital stationar type 1B (GRM6)
2742.	Night blindness, congenital stationar type 1C (TRPM1)
2743.	Night blindness, congenital stationary type 1A (NYX)
2744.	Night blindness, congenital stationary type 2A (CACNA1F)
2745.	Night blindness, congenital stationary type 2B (CABP4)
2746.	Night blindness, congenital stationary type 3 (GNAT1)
2747.	Night blindness, congenital stationary, autosomal dominant type 2 (PDE6B)
2748.	Night blindness, congenital stationary, type 1E (GPR179)
2749.	Nijmegen breakage syndrome (NBN)
2750.	Nonaka myopathy (GNE)

2751.	Nonarteritic anterior ischemic optic neuropathy (GP1BA)
2752.	Nonpolyposis hereditary colon cancer, PMS1 related (PMS1)
2753.	Nonsmall cell lung cancer, familial, susceptibility to (EGFR)
2754.	Nonsmall cell lung cancer, responsive to tyrosin kinase inhibitor, somatic, EGFR related (EGFR)
2755.	Nonsmall cell lung cancer, somatic (BRAF)
2756.	Noonan syndrom like (SHOC2)
2757.	Noonan syndrome type 1 (PTPN11)
2758.	Noonan syndrome type 10 (LZTR1)
2759.	Noonan syndrome type 3 (KRAS)
2760.	Noonan syndrome type 4 (SOS1)
2761.	Noonan syndrome type 5 (RAF1)
2762.	Noonan syndrome type 6 (NRAS)
2763.	Noonan syndrome type 7 (BRAF)
2764.	Noonan syndrome type 8 (RIT1)
2765.	Noonan syndrome-like disorder with or without juvenile meylomonocytic leukemia (CBL)
2766.	Norrie disease (NDP)
2767.	NPM1, selective sequencing of exon 11 (NPM1)
2768.	NRAS somatic Hotspot: c.181C>A p.Q61K (NRAS)
2769.	NRAS somatic Hotspot: c.182A>G, p.Q61R (NRAS)
2770.	NRAS somatic Hotspot: c.34G>T, p.G12C (NRAS)
2771.	NRAS somatic Hotspot: c.35G>A, p.G12D (NRAS)
2772.	NRAS, selective sequencing of exons 2 and 3 (NRAS)
2773.	Nystagmus type 1 (FRMD7)
2774.	Nystagmus type 6 (GPR143)
2775.	Obesity (MC4R)
2776.	Obesity due to leptin deficiency (LEP)
2777.	Obesity with adrenal insufficiency and red hair (POMC)
2778.	Obesity with impaired prohormone processing (PCSK1)
2779.	Obesity, early-onset, susceptibility to (POMC)
2780.	Obesity, severe (PPARG)
2781.	Obesity, susceptibility to, SLC6A14 related (SLC6A14)
2782.	Occipital horn syndrome (ATP7A)

2783.	Occult macular dystrophy (RP1L1)
2784.	Oculodentodigital dysplasia (GJA1)
2785.	Odontohypophosphatasia (ALPL)
2786.	Odontoonychodermal dysplasia (WNT10A)
2787.	Ogden syndrome (NAA10)
2788.	Oguchi disease (SAG)
2789.	Oguchi disease (GRK1)
2790.	Oligo-astheno-teratozoospermia (NANOS1)
2791.	Olmsted syndrome (TRPV3)
2792.	Omenn syndrome (RAG2)
2793.	Omenn syndrome (DCLRE1C)
2794.	Omodysplasia type 1 (GPC6)
2795.	Oocyte maturation defect (ZP1)
2796.	Oogenesis dysfunction (SOHLH1)
2797.	Ophthalmoplegia, isolated, MT-TN related (NGS Sequencing: MT-TN)
2798.	Opitz G syndrome (MID1)
2799.	Opitz-Kaveggia syndrome (MED12)
2800.	Opsismodysplasia (INPPL1)
2801.	Optic atrophy type 1 (OPA1)
2802.	Optic atrophy type 3 (OPA3)
2803.	Optic atrophy type 7 (TMEM126A)
2804.	Optic atrophy type 9 (ACO2)
2805.	Optic atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia, and neuropathy (OPA1)
2806.	Opticoacoustic nerve atrophy with dementia (TIMM8A)
2807.	Oral-facial-digital syndrome type 1 (OFD1)
2808.	Ornithine transcarbamoylase deficiency (OTC)
2809.	Orofacial cleft type 10 (SUMO1)
2810.	Orofacial cleft type 11 (BMP4)
2811.	Orofacial cleft type 5 (MSX1)
2812.	Orofacial cleft type 6 (IRF6)
2813.	Orofacial cleft type 7 (PVRL1)
2814.	Orofaciodigital syndrome type 14 (C2CD3)

2815.	Orofaciodigital syndrome type 4 (TCTN3)
2816.	Orofaciodigital syndrome type 5 (DDX59)
2817.	Orofaciodigital syndrome type 6 (C5orf42)
2818.	Orolaryngeal cancer, multiple, somatic (CDKN2A)
2819.	Orotic aciduria (UMPS)
2820.	Orthostatic intolerance (SLC6A2)
2821.	Osseous heteroplasia, progressive (GNAS)
2822.	Osteoarthritis with mild chondrodysplasia (COL2A1)
2823.	Osteogenesis and dental anomalies, CSF1 related (CSF1)
2824.	Osteogenesis disorders, CREB3L1 related (CREB3L1)
2825.	Osteogenesis imperfecta (COL1A1)
2826.	Osteogenesis imperfecta (COL1A2)
2827.	Osteogenesis imperfecta type 10 (SERPINH1)
2828.	Osteogenesis imperfecta type 11 (FKBP10)
2829.	Osteogenesis imperfecta type 12 (SP7)
2830.	Osteogenesis imperfecta type 13 (BMP1)
2831.	Osteogenesis imperfecta type 14 (TMEM38B)
2832.	Osteogenesis imperfecta type 15 (WNT1)
2833.	Osteogenesis imperfecta type 5 (IFITM5)
2834.	Osteogenesis imperfecta type 6 (SERPINF1)
2835.	Osteogenesis imperfecta type 7 (CRTAP)
2836.	Osteogenesis imperfecta type 8 (P3H1)
2837.	Osteogenesis imperfecta type 9 (PPIB)
2838.	Osteogenesis imperfecta with congenital joint contractures (PLOD2)
2839.	Osteoglophonic dysplasia (FGFR1)
2840.	Osteolysis, familial expansile (TNFRSF11A)
2841.	Osteomyelitis, sterile multifocal, with periostitis and pustulosis (IL1RN)
2842.	Osteopathia striata with cranial sclerosis (AMER1)
2843.	Osteopetrosis of infancy, malignant (SNX10)
2844.	Osteopetrosis, autosomal dominant type 1 (CLCN7)
2845.	Osteopetrosis, autosomal recessive type 1 (TCIRG1)
2846.	Osteopetrosis, autosomal recessive type 2 (TNFSF11)

2847.	Osteopetrosis, autosomal recessive type 3 (CA2)
2848.	Osteopetrosis, autosomal recessive type 4 (CLCN7)
2849.	Osteopetrosis, autosomal recessive type 5 (OSTM1)
2850.	Osteopetrosis, autosomal recessive type 6 (PLEKHM1)
2851.	Osteopetrosis, autosomal recessive type 7 (TNFRSF11A)
2852.	Osteoporosis pseudoglioma syndrome (LRP5)
2853.	Otofaciocervical syndrome (EYA1)
2854.	Otopaladigital syndrome type 1 (FLNA)
2855.	Otopaladigital syndrome type 2 (FLNA)
2856.	Otospondylomegaepiphyseal dysplasia (COL2A1)
2857.	Otospondylomegaepiphyseal dysplasia (COL11A2)
2858.	Ovalocytosis (SLC4A1)
2859.	Ovarian cancer, somatic (CTNNB1)
2860.	Ovarian cancer, somatic (ERBB2)
2861.	Ovarian carcinoma, somatic (CDH1)
2862.	Ovarian clear-cell carcinoma, ARID1A related, somatic (ARID1A)
2863.	Ovarian dysgenesis type 1 (FSHR)
2864.	Ovarian dysgenesis type 2 (BMP15)
2865.	Pachyonychia congenita type 1 (KRT16)
2866.	Pachyonychia congenita type 2 (KRT17)
2867.	Pachyonychia congenita type 3 (KRT6A)
2868.	Pachyonychia congenita type 4 (KRT6B)
2869.	Paget disease of bone (SQSTM1)
2870.	Paget disease, juvenile (TNFRSF11B)
2871.	Pallister-Hall syndrome (GLI3)
2872.	Palmoplantar keratoderma, nonepidermolytic, focal (KRT16)
2873.	Pancreatic agenesis and congenital heart defects (GATA6)
2874.	Pancreatic agenesis type 2 (PTF1A)
2875.	Pancreatic and cerebellar agenesis (PTF1A)
2876.	Pancreatic cancer type 2, susceptibility to (BRCA2)
2877.	Pancreatic cancer type 3, susceptibility to (PALB2)
2878.	Pancreatic cancer, somatic (SMAD4)

2879.	Pancreatic cancer, somatic (STK11)
2880.	Pancreatic cancer, somatic (TP53)
2881.	Pancreatic cancer, susceptibility to, type 4 (BRCA1)
2882.	Pancreatic cancer/melanoma syndrome, familial (CDKN2A)
2883.	Pancreatic carcinoma, somatic (KRAS)
2884.	Pancreatitis (CTRC)
2885.	Pancreatitis (SPINK1)
2886.	Pancreatitis (PRSS1)
2887.	Pancreatitis, chronic, early onset (CPA1)
2888.	Pancreatitis, chronic, protection against (PRSS2)
2889.	Panhypopituitarism, X-linked (SOX3)
2890.	Pantothenate kinase-associated neurodegeneration (PANK2)
2891.	Papillon-Lefevre syndrome (CTSC)
2892.	Papillorenal syndrome (PAX2)
2893.	Paraganglioma and gastric stromal sarcoma (SDHD)
2894.	Paragangliomas type 1, with or without deafness (SDHD)
2895.	Paragangliomas type 4 (SDHB)
2896.	Paragangliomas type 5 (SDHA)
2897.	Paramyotonia congenita of von Eulenburg (SCN4A)
2898.	Parathyroid adenoma with cystic changes, familial (CDC73)
2899.	Parathyroid adenoma, somatic (MEN1)
2900.	Parietal foramina type 1 (MSX2)
2901.	Parietal foramina type 2 (ALX4)
2902.	PARK1 Parkinson (SNCA)
2903.	PARK13 Parkinson (HTRA2)
2904.	PARK14 Parkinson (PLA2G6)
2905.	PARK15 Parkinson (FBXO7)
2906.	PARK17 Parkinson (VPS35)
2907.	PARK19 Parkinson, juvenile-onset (DNAJC6)
2908.	PARK2 Parkinson (PARK2)
2909.	PARK20 Parkinson (SYNJ1)
2910.	PARK21 Parkinson (DNAJC13)

2911.	PARK4 Parkinson (SNCA)
2912.	PARK5 Parkinson (UCHL1)
2913.	PARK6 Parkinson (PINK1)
2914.	PARK7 Parkinson (PARK7)
2915.	PARK8 Parkinson (LRRK2)
2916.	PARK9 Parkinson (ATP13A2)
2917.	Parkes Weber syndrome (RASA1)
2918.	Parkinson disease, late-onset, susceptibility to (GBA)
2919.	Parkinson disease, susceptibility to, MT-TT related (NGS Sequencing: MT-TT)
2920.	Parkinsonism with spasticity, X-linked (ATP6AP2)
2921.	Parkinsonism-Dystonia, infantile (SLC6A3)
2922.	Paroxysmal exercise-induced dyskinesia with epilepsy and/or hemolytic anemia (SLC2A1)
2923.	Paroxysmal nonkinesigenic dyskinesia (PNKD)
2924.	Partington syndrome (ARX)
2925.	Patterned dystrophy of retinal pigment epithelium (PRPH2)
2926.	PDGFRA, selective sequencing of exons 12, 14 and 18 (PDGFRA)
2927.	Peeling skin syndrome type 1 (CDSN)
2928.	Peeling skin syndrome type 2 (TGM5)
2929.	Peeling skin syndrome type 3 (CHST8)
2930.	Peeling skin syndrome type 4 (CSTA)
2931.	Pelger-Huet anomaly (LBR)
2932.	Pelizaeus-Merzbacher disease (SLC16A2)
2933.	Pelizaeus-Merzbacher disease (PLP1)
2934.	Pelvic organ prolapse, LAMC1 related (LAMC1)
2935.	Pendred syndrome (SLC26A4)
2936.	Pentosuria (DCXR)
2937.	Periodic fever autosomal dominant (TNFRSF1A)
2938.	Peripheral demyelinating neuropathy Waardenburg syndrome and Hirschsprung disease (SOX10)
2939.	Periventricular heterotopia with microcephaly (ARFGEF2)
2940.	Perlman Syndrome (DIS3L2)
2941.	Peroxisome biogenesis disorder 14B (PEX11B)
2942.	Peroxisome biogenesis disorder type 10A (PEX3)

2943.	Peroxisome biogenesis disorder type 1B (PEX1)
2944.	Peroxisome biogenesis disorder type 2A (PEX5)
2945.	Peroxisome biogenesis disorder type 2B (PEX5)
2946.	Perrault syndrome (HSD17B4)
2947.	Perrault syndrome type 5 (TWNK)
2948.	Persistent Mullerian duct syndrome type 1 (AMH)
2949.	Persistent Mullerian duct syndrome type 2 (AMHR2)
2950.	Peters Anomaly (PITX2)
2951.	Peters anomaly (PAX6)
2952.	Peters Anomaly (CYP1B1)
2953.	Peters-Plus syndrome (B3GLCT)
2954.	Peutz-Jeghers syndrome (STK11)
2955.	Peutz-Jeghers syndrome, somatic (STK11)
2956.	Pfeiffer syndrome (FGFR1)
2957.	Pfeiffer syndrome (FGFR2)
2958.	Phelan-McDermid syndrome (SHANK3)
2959.	Phelan-McDermid syndrome (chr. 22q13.3)
2960.	Phenylketonuria (PAH)
2961.	Phenylketonuria modifier, SLC7A5 related (SLC7A5)
2962.	Pheochromocytoma type 1 (SDHD)
2963.	Pheochromocytoma type 2 (SDHB)
2964.	Pheochromocytoma type 3 (SDHC)
2965.	Pheochromocytoma type 5 (SDHAF2)
2966.	Pheochromocytoma type 8 (TMEM127)
2967.	Pheochromocytoma type 9 (MAX)
2968.	Phosphoenolpyruvate carboxykinase deficiency, cytosolic (PCK1)
2969.	Phosphoenolpyruvate carboxykinase deficiency, mitochondrial (PCK2)
2970.	Phosphoglycerate dehydrogenase deficiency (PHGDH)
2971.	Phosphoglycerate kinase 1 deficiency (PGK1)
2972.	Phosphoribosylpyrophosphate synthetase superactivity (PRPS1)
2973.	Phosphoserine aminotransferase deficiency (PSAT1)
2974.	Phosphoserine phosphatase deficiency (PSPH)

2975.	Pick disease (PSEN1)
2976.	Piebaldism (KIT)
2977.	Piebaldism (SNAI2)
2978.	Pigmented nodular adrenocortical disease type 1, primary (PRKAR1A)
2979.	Pigmented paravenous chorioretinal atrophy (CRB1)
2980.	PIK3CA related overgrowth spectrum, somatic (PIK3CA)
2981.	Pitt-Hopkins syndrome (NRXN1)
2982.	Pitt-Hopkins syndrome (TCF4)
2983.	Pituitary adenoma, ACTH-secreting, due to AIP germline mutation (AIP)
2984.	Pituitary adenoma, growth hormone-secreting, due to AIP germline mutation (AIP)
2985.	Pituitary adenoma, prolactin-secreting, due to AIP germline mutation (AIP)
2986.	Pituitary hormone deficiency type 1 (POU1F1)
2987.	Pituitary hormone deficiency type 2 (PROP1)
2988.	Pituitary hormone deficiency, combined type 3 (LHX3)
2989.	Pituitary hormone deficiency, combined type 4 (LHX4)
2990.	Pituitary stalk interruption syndrome, GPR161 related (GPR161)
2991.	Pityriasis rubra pilaris (CARD14)
2992.	Plasminogen activator inhibitor type 1 (SERPINE1)
2993.	Plasminogen deficiency type 1 (PLG)
2994.	Platelet aggregation disorder (PEAR1)
2995.	Platelet dense granule secretion defect, excessive bleeding (FLI1)
2996.	Platelet disorder with associated myeloid malignancy (RUNX1)
2997.	Platelet glycoprotein IV deficiency (CD36)
2998.	Platyspondylic skeletal dysplasia, Torrance type (COL2A1)
2999.	Pleuropulmonary blastoma (DICER1)
3000.	Pneumothorax, primary spontaneous (FLCN)
3001.	Poikiloderma with neutropenia (USB1)
3002.	Polyarteritis nodosa, childhood-onset (CECR1)
3003.	Polycystic kidney and hepatic disease (NGS Sequencing: PKHD1)
3004.	Polycystic kidney disease type 1, autosomal dominant (PKD1)
3005.	Polycystic kidney disease type 1, autosomal recessive (NGS Sequencing: PKHD1)
3006.	Polycystic kidney disease type 2, autosomal dominant (PKD2)

3007.	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (TREM2)
3008.	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (TYROBP)
3009.	Polycystic liver disease (PRKCSH)
3010.	Polycystic ovary syndrome type 1 (SULT2B1)
3011.	Polycystic ovary syndrome type 1 (SULT2A1)
3012.	Polycythemia vera, somatic (JAK2)
3013.	Polyglucosan body myopathy type 1 with or without immunodeficiency (RBCK1)
3014.	Polymicrogyria asymmetric (TUBB2B)
3015.	Polymicrogyria bilateral frontoparietal (ADGRG1)
3016.	Polymicrogyria bilateral occipital (NR2E1)
3017.	Polymicrogyria with optic nerve hypoplasia (TUBA8)
3018.	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis (PI4KA)
3019.	Polyposis syndrome, hereditary mixed (GREM1)
3020.	Polyposis syndrome, hereditary mixed type 2 (BMPR1A)
3021.	Pompe disease (GAA)
3022.	Pontocerebellar hypoplasia type 1A (VRK1)
3023.	Pontocerebellar hypoplasia type 1B (EXOSC3)
3024.	Pontocerebellar hypoplasia type 2A (TSEN54)
3025.	Pontocerebellar hypoplasia type 2B (TSEN2)
3026.	Pontocerebellar hypoplasia type 2C (TSEN34)
3027.	Pontocerebellar hypoplasia type 2D (SEPSECS)
3028.	Pontocerebellar hypoplasia type 2E (VPS53)
3029.	Pontocerebellar hypoplasia type 4 (TSEN54)
3030.	Pontocerebellar hypoplasia type 5 (TSEN54)
3031.	Pontocerebellar hypoplasia type 6 (RARS2)
3032.	Pontocerebellar hypoplasia type 8 (CHMP1A)
3033.	Pontocerebellar hypoplasia, type 10 (CLP1)
3034.	Pontocerebellar hypoplasia, type 9 (AMPD2)
3035.	Popliteal pterygium syndrome type 1 (IRF6)
3036.	Popliteal pterygium syndrome, lethal type (RIPK4)
3037.	Porencephaly type 2 (COL4A2)
3038.	Porencephaly, familial (COL4A1)

3039.	Poretti-Boltshauser syndrome (NGS Sequencing: LAMA1)
3040.	Porokeratosis type 3, disseminated superficial actinic (MVK)
3041.	Porphyria acute intermittent (HMBS)
3042.	Porphyria congenital erythropoietic (UROS)
3043.	Porphyria cutanea tarda (UROD)
3044.	Porphyria variegata (PPOX)
3045.	Postaxial acrofacial dysostosis (DHODH)
3046.	Potassium-aggravated myotonia (SCN4A)
3047.	Prader-Willi syndrome (SNRPN)
3048.	Prader-Willi syndrome (NDN)
3049.	Prader-Willi syndrome (chr. 15q11)
3050.	Precocious puberty, male (LHCGR)
3051.	Preeclampsia/eclampsia type 5 (CORIN)
3052.	Pregnancy loss, recurrent, C4BPA related (C4BPA)
3053.	Premature ovarian failure type 1 (FMR1)
3054.	Primary aldosteronism, seizures, and neurologic abnormalities (CACNA1D)
3055.	Primary ciliary dyskinesia type 1 (DNAI1)
3056.	Primary ciliary dyskinesia type 10 (DNAAF2)
3057.	Primary ciliary dyskinesia type 11 (RSPH4A)
3058.	Primary ciliary dyskinesia type 12 (RSPH9)
3059.	Primary ciliary dyskinesia type 13 (DNAAF1)
3060.	Primary ciliary dyskinesia type 14 (CCDC39)
3061.	Primary ciliary dyskinesia type 15 (CCDC40)
3062.	Primary ciliary dyskinesia type 16 (DNAL1)
3063.	Primary ciliary dyskinesia type 17 (CCDC103)
3064.	Primary ciliary dyskinesia type 18 (DNAAF5)
3065.	Primary ciliary dyskinesia type 19 (LRRC6)
3066.	Primary ciliary dyskinesia type 2 (DNAAF3)
3067.	Primary ciliary dyskinesia type 20 (CCDC114)
3068.	Primary ciliary dyskinesia type 23 (ARMC4)
3069.	Primary ciliary dyskinesia type 24 (RSPH1)
3070.	Primary ciliary dyskinesia type 25 (DYX1C1)

3071.	Primary ciliary dyskinesia type 26 (C21orf59)
3072.	Primary ciliary dyskinesia type 27 (CCDC65)
3073.	Primary ciliary dyskinesia type 28 (SPAG1)
3074.	Primary ciliary dyskinesia type 29 (CCNO)
3075.	Primary ciliary dyskinesia type 3 (NGS Sequencing: DNAH5)
3076.	Primary ciliary dyskinesia type 5 (NGS Sequencing: HYDIN)
3077.	Primary ciliary dyskinesia type 6 (NME8)
3078.	Primary ciliary dyskinesia type 7 (NGS Sequencing: DNAH11)
3079.	Primary ciliary dyskinesia type 9 (DNAI2)
3080.	Primary ciliary dyskinesia, DNAH9 related (NGS Sequencing: DNAH9)
3081.	Primary lateral sclerosis, juvenile (ALS2)
3082.	Progressive external ophthalmoplegia with mitochondrial deletions type 1 (POLG)
3083.	Progressive external ophthalmoplegia with mitochondrial deletions type 2 (SLC25A4)
3084.	Progressive external ophthalmoplegia with mitochondrial deletions type 3 (TWNK)
3085.	Progressive external ophthalmoplegia with mitochondrial deletions type 4 (POLG2)
3086.	Progressive external ophthalmoplegia with mitochondrial deletions type 5 (RRM2B)
3087.	Progressive external ophthalmoplegia with mitochondrial deletions type 6 (DNA2)
3088.	Progressive external ophthalmoplegia with mitochondrial deletions, autosomal recessive (POLG)
3089.	Progressive familial heart block (TRPM4)
3090.	Progressive hearing loss (P2RX2)
3091.	Progressive myoclonus epilepsy type 1A (PRICKLE1)
3092.	Progressive myoclonus epilepsy type 3 (KCTD7)
3093.	Progressive myoclonus epilepsy type 6 (GOSR2)
3094.	Progressive myoclonus epilepsy type 8 (CERS1)
3095.	Prolidase deficiency (PEPD)
3096.	Propionic acidemia (PCCA)
3097.	Propionic acidemia (PCCB)
3098.	Prosaposin deficiency (PSAP)
3099.	Prostate cancer (ZNF783)
3100.	Prostate cancer (SRD5A2)
3101.	Prostate cancer (STAG1)
3102.	Prostate cancer (BRCA2)

3103.	Prostate cancer, familial, association with (HOXB13)
3104.	Prostate cancer, hereditary type 1 (RNASEL)
3105.	Prostate cancer, hereditary type 2, susceptibility to (ELAC2)
3106.	Prostate cancer, somatic (KLF6)
3107.	Prostate tumor, AR related, somatic (AR)
3108.	Protein C Deficiency, AD (PROC)
3109.	Protein S Deficiency, autosomal dominant (PROS1)
3110.	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis (CLCN5)
3111.	Protoporphyria, erythropoietic, X-linked (ALAS2)
3112.	Prune belly syndrome (CHRM3)
3113.	Pseudoachondroplasia (COMP)
3114.	Pseudohermaphroditism with gynecomastia (HSD17B3)
3115.	Pseudohypoaldosteronism type 2D (KLHL3)
3116.	Pseudohypoaldosteronism type 2E (CUL3)
3117.	Pseudohypoaldosteronism, type 1, autosomal dominant (NR3C2)
3118.	Pseudohypoaldosteronism, type 1, autosomal recessive (SCNN1B)
3119.	Pseudohypoaldosteronism, type 1, autosomal recessive (SCNN1G)
3120.	Pseudohypoaldosteronism, type 1, autosomal recessive (SCNN1A)
3121.	Pseudohypoaldosteronism, type 2B (WNK4)
3122.	Pseudohypoparathyroidism type 1A (GNAS)
3123.	Pseudohypoparathyroidism type 1B (GNAS)
3124.	Pseudohypoparathyroidism type 1C (GNAS)
3125.	Pseudopseudohypoparathyroidism (GNAS)
3126.	Pseudoxanthoma elasticum (ABCC6)
3127.	Pseudoxanthoma elasticum, forme fruste (ABCC6)
3128.	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency (GGCX)
3129.	Psoriasis susceptibility type 11 (IL12B)
3130.	Psoriasis type 2 (CARD14)
3131.	Psoriasis, generalized pustular (IL36RN)
3132.	Psychomotor retardation (TANC1)
3133.	Pterygium syndrome (CHRNG)
3134.	Ptosis, congenital (ZFHX4)

3135.	Pulmonary fibrosis and/or bone marrow failure, telomere-related, type 1 (TERT)
3136.	Pulmonary fibrosis and/or bone marrow failure, telomere-related, type 4 (PARN)
3137.	Pulmonary fibrosis, idiopathic (SFTPA2)
3138.	Pulmonary fibrosis, idiopathic (SFTPA1)
3139.	Pulmonary hypertension, primary type (BMPR2)
3140.	Pulmonary newborn hypertension (CRHR1)
3141.	Pulmonary venoocclusive disease type 1 (BMPR2)
3142.	Pulmonary venoocclusive disease type 2 (EIF2AK4)
3143.	Pycnodysostosis (CTSK)
3144.	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency (MYD88)
3145.	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne (PSTPIP1)
3146.	Pyridoxamine 5'-phosphate oxidase deficiency (PNPO)
3147.	Pyridoxine-dependent epilepsy (ALDH7A1)
3148.	Pyruvate carboxylase deficiency (PC)
3149.	Pyruvate dehydrogenase E1-alpha deficiency (PDHA1)
3150.	Pyruvate dehydrogenase E1-beta deficiency (PDHB)
3151.	Pyruvate dehydrogenase E2 deficiency (DLAT)
3152.	Pyruvate dehydrogenase lipoic acid synthetase deficiency (LIAS)
3153.	Pyruvate dehydrogenase phosphatase deficiency (PDP1)
3154.	Pyruvate kinase deficiency with hemolytic anemia (PKLR)
3155.	Radioulnar synostosis, FGFRL1 related (FGFRL1)
3156.	Raine syndrome (FAM20C)
3157.	RAPADILINO syndrome (RECQL4)
3158.	Rapp-Hodgkin syndrome (TP63)
3159.	Refsum disease (PHYH)
3160.	Refsum disease (PEX7)
3161.	Renal cancer, KDM6A related, somatic (KDM6A)
3162.	Renal carcinoma, chromophobe, somatic (FLCN)
3163.	Renal carcinoma, Tuberous sclerosis-associated, somatic (TSC1)
3164.	Renal cell carcinoma, due to HNF1A germline mutation (HNF1A)
3165.	Renal cell carcinoma, papillary type 1, familial (MET)
3166.	Renal cell carcinoma, papillary type 1, somatic (MET)

3167.	Renal cell carcinoma, somatic (VHL)
3168.	Renal cystic dysplasia, cystic, susceptibility to (BICC1)
3169.	Renal dysfunction due to SLC26A1 deficiency (SLC26A1)
3170.	Renal glucosuria (SLC5A2)
3171.	Renal tubular acidosis with deafness (ATP6V1B1)
3172.	Renal tubular acidosis, distal, autosomal recessive (ATP6V0A4)
3173.	Renal tubular acidosis, proximal, with ocular abnormalities (SLC4A4)
3174.	Renal tubular acidosis, SLC4A5 related (SLC4A5)
3175.	Renal tubular dysgenesis (ACE)
3176.	Renal tubular dysgenesis (AGT)
3177.	Renal tubular dysgenesis (REN)
3178.	Renal tubular dysgenesis (AGTR1)
3179.	Renpenning syndrome (PQBP1)
3180.	Restrictive dermopathy, lethal (LMNA)
3181.	Restrictive dermopathy, lethal (ZMPSTE24)
3182.	RET, selective sequencing of exons 5, 8, 10, 11 and 13-16 (RET)
3183.	Reticular dysgenesis (AK2)
3184.	Retinal cone dystrophy type 3B (KCNV2)
3185.	Retinal cone dystrophy type 4 (CACNA2D4)
3186.	Retinal degeneration, late-onset, autosomal dominant (C1QTNF5)
3187.	Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities (ITM2B)
3188.	Retinal nonattachment nonsyndromic congenital (ATOH7)
3189.	Retinitis pigmentosa juvenile (LRAT)
3190.	Retinitis pigmentosa SEMA4C related (SEMA4C)
3191.	Retinitis pigmentosa type 1, autosomal dominant (RP1)
3192.	Retinitis pigmentosa type 10, autosomal dominant (IMPDH1)
3193.	Retinitis pigmentosa type 11, autosomal dominant (PRPF31)
3194.	Retinitis pigmentosa type 12, autosomal recessive (CRB1)
3195.	Retinitis pigmentosa type 13, autosomal dominant (PRPF8)
3196.	Retinitis pigmentosa type 14, autosomal recessive (TULP1)
3197.	Retinitis pigmentosa type 17, autosomal dominant (CA4)
3198.	Retinitis pigmentosa type 18, autosomal dominant (PRPF3)

3199.	Retinitis pigmentosa type 19, autosomal recessive (ABCA4)
3200.	Retinitis pigmentosa type 2 X-linked (RP2)
3201.	Retinitis pigmentosa type 20, autosomal recessive (RPE65)
3202.	Retinitis pigmentosa type 23 X-linked (OFD1)
3203.	Retinitis pigmentosa type 25 (NGS Sequencing: EYS)
3204.	Retinitis pigmentosa type 26, autosomal recessive (CERKL)
3205.	Retinitis pigmentosa type 27, autosomal dominant (NRL)
3206.	Retinitis pigmentosa type 28, autosomal recessive (FAM161A)
3207.	Retinitis pigmentosa type 3 X-linked (RPGR)
3208.	Retinitis pigmentosa type 30, autosomal dominant (FSCN2)
3209.	Retinitis pigmentosa type 31, autosomal dominant (TOPORS)
3210.	Retinitis pigmentosa type 33, autosomal dominant (SNRNP200)
3211.	Retinitis pigmentosa type 35, autosomal dominant/recessive (SEMA4A)
3212.	Retinitis pigmentosa type 36, autosomal recessive (PRCD)
3213.	Retinitis pigmentosa type 37, autosomal dominant/recessive (NR2E3)
3214.	Retinitis pigmentosa type 38, autosomal recessive (MERTK)
3215.	Retinitis pigmentosa type 39 (NGS Sequencing: USH2A)
3216.	Retinitis pigmentosa type 4, autosomal dominant/recessive (RHO)
3217.	Retinitis pigmentosa type 40, autosomal recessive (PDE6B)
3218.	Retinitis pigmentosa type 41, autosomal recessive (PROM1)
3219.	Retinitis pigmentosa type 42, autosomal dominant (KLHL7)
3220.	Retinitis pigmentosa type 43, autosomal recessive (PDE6A)
3221.	Retinitis pigmentosa type 44, autosomal dominant/recessive (RGR)
3222.	Retinitis pigmentosa type 45, autosomal recessive (CNGB1)
3223.	Retinitis pigmentosa type 46, autosomal recessive (IDH3B)
3224.	Retinitis pigmentosa type 47, autosomal recessive (SAG)
3225.	Retinitis pigmentosa type 48, autosomal dominant (GUCA1B)
3226.	Retinitis pigmentosa type 49, autosomal recessive (CNGA1)
3227.	Retinitis pigmentosa type 50, autosomal dominant (BEST1)
3228.	Retinitis pigmentosa type 51, autosomal recessive (TTC8)
3229.	Retinitis pigmentosa type 53, autosomal recessive (RDH12)
3230.	Retinitis pigmentosa type 54, autosomal recessive (C2ORF71)

3231.	Retinitis pigmentosa type 55, autosomal recessive (ARL6)
3232.	Retinitis pigmentosa type 56, autosomal recessive (IMPG2)
3233.	Retinitis pigmentosa type 57, autosomal recessive (PDE6G)
3234.	Retinitis pigmentosa type 58, autosomal recessive (ZNF513)
3235.	Retinitis pigmentosa type 59, autosomal recessive (DHDDS)
3236.	Retinitis pigmentosa type 60 (PRPF6)
3237.	Retinitis pigmentosa type 61, autosomal recessive (CLRN1)
3238.	Retinitis pigmentosa type 62, autosomal recessive (MAK)
3239.	Retinitis pigmentosa type 64, autosomal recessive (C8ORF37)
3240.	Retinitis pigmentosa type 66, autosomal recessive (RBP3)
3241.	Retinitis pigmentosa type 7 (ROM1)
3242.	Retinitis pigmentosa type 7, autosomal dominant (PRPH2)
3243.	Retinitis pigmentosa type 74, autosomal recessive (BBS2)
3244.	Retinitis pigmentosa type 9, autosomal dominant (RP9)
3245.	Retinitis pigmentosa, juvenile, autosomal recessive (SPATA7)
3246.	Retinitis punctata albescens (RHO)
3247.	Retinoschisis (RS1)
3248.	Rett syndrome (MECP2)
3249.	Rett syndrome preserved speech variant (MECP2)
3250.	Rett syndrome, congenital variant (FOXG1)
3251.	Revesz syndrome (TINF2)
3252.	Rhabdoid tumors, somatic (SMARCB1)
3253.	Rheumatoid arthritis, susceptibility to (AFF3)
3254.	Rheumatoid arthritis, TNFAIP3 related (TNFAIP3)
3255.	Rhizomelic chondrodysplasia punctata type 2 (GNPAT)
3256.	Rhizomelic chondrodysplasia punctata type 3 (AGPS)
3257.	Rhizomelic chondrodysplasia punctata type 5 (PEX5)
3258.	Riboflavin deficiency (SLC52A1)
3259.	Rickets, vitamin D 25-hydroxylation-deficient, type 1B (CYP2R1)
3260.	Rickets, vitamin D dependent, type 1 (CYP27B1)
3261.	Rickets, vitamin D-resistant, type 2A (VDR)
3262.	Rigid spine muscular dystrophy (SELENON)

3263.	Ring dermoid of cornea (PITX2)
3264.	Rippling muscle disease (CAV3)
3265.	Ritscher-Schinzel syndrome type 1 (KIAA0196)
3266.	RNA processing related disorders (HNRNPU)
3267.	Roberts syndrome (ESCO2)
3268.	Robinow syndrome, autosomal dominant type 1 (WNT5A)
3269.	Robinow syndrome, autosomal dominant type 2 (DVL1)
3270.	Robinow syndrome, autosomal recessive (ROR2)
3271.	Robinow-Sorauf syndrome (TWIST1)
3272.	Rolandic epilepsy, mental retardation, and speech dyspraxia (SRPX2)
3273.	Rothmund-Thomson syndrome (RECQL4)
3274.	Roussy-Levy syndrome (PMP22)
3275.	Rubinstein-Taybi syndrome (CREBBP)
3276.	Rubinstein-Taybi syndrome (EP300)
3277.	Saccharopinuria (AASS)
3278.	Saethre-Chotzen syndrome (TWIST1)
3279.	Saethre-Chotzen syndrome (FGFR2)
3280.	Salih ataxia (RUBCN)
3281.	Sandhoff disease (HEXB)
3282.	Sarcoidosis, early-onset (NOD2)
3283.	Sarcosinemia (SARDH)
3284.	SC Phocomelia syndrome (ESCO2)
3285.	Scaphocephaly, maxillary retrusion, and mental retardation (FGFR2)
3286.	Scapuloperoneal myopathy, MYH7 related (MYH7)
3287.	Schaaf-Yang syndrome (MAGEL2)
3288.	Scheie syndrome (IDUA)
3289.	Schimke immunoosseous dysplasia (SMARCAL1)
3290.	Schindler disease (NAGA)
3291.	Schinzel-Giedion midface retraction syndrome (SETBP1)
3292.	Schizencephaly (EMX2)
3293.	Schizophrenia, CALR related (CALR)
3294.	Schizophrenia, CELSR2 related (CELSR2)

3295.	Schizophrenia, GRID2 related (GRID2)
3296.	Schizophrenia, NOTCH4 related (NOTCH4)
3297.	Schneckenbecken dysplasia (SLC35D1)
3298.	Schwartz-Jampel syndrome type 1 (NGS Sequencing: HSPG2)
3299.	SCID autosomal recessive T negative B positive type (JAK3)
3300.	Sclerosteosis type 1 (SOST)
3301.	Sea-blue histiocyte disease (APOE)
3302.	Seckel syndrome (ATRIP)
3303.	Seckel syndrome type 1 (NGS Sequencing: ATR)
3304.	Seckel syndrome type 2 (RBBP8)
3305.	Seckel syndrome type 4 (CENPJ)
3306.	Seckel syndrome type 5 (CEP152)
3307.	Seckel syndrome type 6 (CEP63)
3308.	Seckel syndrome type 7 (NIN)
3309.	SED congenita (COL2A1)
3310.	Segawa syndrome, autosomal recessive (TH)
3311.	Seizures, benign familial infantile, type 2 (PRRT2)
3312.	Seizures, benign neonatal, type 1 (KCNQ2)
3313.	Seizures, benign neonatal, type 2 (KCNQ3)
3314.	Seizures, scoliosis, and macrocephaly syndrome (EXT2)
3315.	Selective T-cell defect (ZAP70)
3316.	Sengers syndrome (AGK)
3317.	Senior-Loken syndrome type 5 (IQCB1)
3318.	Senior-Loken syndrome type 6 (CEP290)
3319.	Senior-Loken syndrome type 7 (SDCCAG8)
3320.	Senior-Loken syndrome type 8 (WDR19)
3321.	Septooptic dysplasia (HESX1)
3322.	Serine hydrolase deficiency, SERHL2 related (SERHL2)
3323.	SERKAL syndrome (WNT4)
3324.	SESAME syndrome (KCNJ10)
3325.	Severe combined immunodeficiency due to ADA deficiency (ADA)
3326.	Severe combined immunodeficiency due to IL2 deficiency (IL2)

3327.	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation (NHEJ1)
3328.	Severe combined immunodeficiency, Athabascan type (DCLRE1C)
3329.	Severe combined immunodeficiency, B cell-negative (RAG1)
3330.	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive (PTPRC)
3331.	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type (IL7R)
3332.	Short QT syndrome type 1 (KCNH2)
3333.	Short QT syndrome type 2 (KCNQ1)
3334.	Short QT syndrome type 3 (KCNJ2)
3335.	Short stature syndrome (SHOX)
3336.	Short stature, microcephaly, and endocrine dysfunction (XRCC4)
3337.	Short stature, optic nerve atrophy, and Pelger-Huet anomaly (NBAS)
3338.	SHORT syndrome (PIK3R1)
3339.	Short-rib thoracic dysplasia type 10 with or without polydactyly (IFT172)
3340.	Short-rib thoracic dysplasia type 11 with or without polydactyly (WDR34)
3341.	Short-rib thoracic dysplasia type 2 with or without polydactyly (IFT80)
3342.	Short-rib thoracic dysplasia type 3 with or without polydactyly (NGS Sequencing: DYNC2H1)
3343.	Short-rib thoracic dysplasia type 4 with or without polydactyly (TTC21B)
3344.	Short-rib thoracic dysplasia type 5 with or without polydactyly (WDR19)
3345.	Short-rib thoracic dysplasia type 6 with or without polydactyly (NEK1)
3346.	Short-rib thoracic dysplasia type 7 with or without polydactyly (WDR35)
3347.	Short-rib thoracic dysplasia type 8 with or without polydactyly (WDR60)
3348.	Shprintzen-Goldberg syndrome (SKI)
3349.	Shwachman-Diamond syndrome (SBDS)
3350.	Sialuria, finish type (SLC17A5)
3351.	Sick sinus syndrome type 1 (SCN5A)
3352.	Sick sinus syndrome type 3 (MYH6)
3353.	Sickle cell anemia (HBB)
3354.	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (TRNT1)
3355.	Silver-Russell syndrome (chr. 11p15)
3356.	Silver-Russell syndrome (maternal UPD chr. 7)
3357.	Simpson-Golabi-Behmel syndrome type 1 (GPC3)
3358.	Sinoatrial node dysfunction and deafness (CACNA1D)

3359.	Sjogren-Larsson syndrome (ALDH3A2)
3360.	Skeletal abnormalities, CBFB related (CBFB)
3361.	Skin fragility-woolly hair syndrome (DSP)
3362.	Skin hair eye pigmentation type 6 (SLC24A4)
3363.	Slowed nerve conduction velocity, autosomanal dominant (ARHGEF10)
3364.	Small cell ovarian carcinoma, hypercalcemic type, SMARCA4 related, somatic (SMARCA4)
3365.	SMED Strudwick type (COL2A1)
3366.	Smith-Lemli-Opitz syndrome (DHCR7)
3367.	Smith-Magenis syndrome (RAI1)
3368.	Smith-Magenis syndrome, ULK2 related (ULK2)
3369.	Smith-McCort dysplasia (DYM)
3370.	Sneddon syndrome (CECR1)
3371.	Sorsby fundus dystrophy (TIMP3)
3372.	Sotos syndrome type 1 (NSD1)
3373.	Sotos-like syndrome (NFIX)
3374.	Spastic ataxia Charlevoix-Saguenay type (SACS)
3375.	Spastic ataxia type 1, autosomal dominant (VAMP1)
3376.	Spastic ataxia type 2, autosomal recessive (KIF1C)
3377.	Spastic ataxia type 3, autosomal recessive (MARS2)
3378.	Spastic ataxia type 5, autosomal recessive (AFG3L2)
3379.	Spastic paralysis, infantile onset ascending (ALS2)
3380.	Spastic paraplegia type 74, autosomal recessive (IBA57)
3381.	Speech-language disorder type 1 (FOXP2)
3382.	Spermatocytic seminoma, somatic (FGFR3)
3383.	SPG1 (L1CAM)
3384.	SPG10 (KIF5A)
3385.	SPG11 (SPG11)
3386.	SPG12 (RTN2)
3387.	SPG13 (HSPD1)
3388.	SPG15 (ZFYVE26)
3389.	SPG17 (BSCL2)
3390.	SPG18 (ERLIN2)

3391.	SPG2 (PLP1)
3392.	SPG20 (SPG20)
3393.	SPG21 (SPG21)
3394.	SPG26 (B4GALNT1)
3395.	SPG28 (DDHD1)
3396.	SPG30 (KIF1A)
3397.	SPG31 (REEP1)
3398.	SPG33 (ZFYVE27)
3399.	SPG35 (FA2H)
3400.	SPG39 (PNPLA6)
3401.	SPG3A (ATL1)
3402.	SPG4 (SPAST)
3403.	SPG42 (SLC33A1)
3404.	SPG43 (C19orf12)
3405.	SPG44 (GJC2)
3406.	SPG45 (NT5C2)
3407.	SPG47 (AP4B1)
3408.	SPG48 (AP5Z1)
3409.	SPG49 (TECPR2)
3410.	SPG50 (AP4M1)
3411.	SPG51 (AP4E1)
3412.	SPG52 (AP4S1)
3413.	SPG53 (VPS37A)
3414.	SPG54 (DDHD2)
3415.	SPG55 (C12ORF65)
3416.	SPG56 (CYP2U1)
3417.	SPG57 (TFG)
3418.	SPG59, USP8 related (USP8)
3419.	SPG5A (CYP7B1)
3420.	SPG6 (NIPA1)
3421.	SPG60, WDR48 related (WDR48)
3422.	SPG61 (ARL6IP1)

3423.	SPG62, ERLIN1 related (ERLIN1)
3424.	SPG63 (AMPD2)
3425.	SPG64 (ENTPD1)
3426.	SPG66, ARSI related (ARSI)
3427.	SPG68, FLRT1 related (FLRT1)
3428.	SPG7 (SPG7)
3429.	SPG71, ZFR related (ZFR)
3430.	SPG72 (REEP2)
3431.	SPG73 (CPT1C)
3432.	SPG8 (KIAA0196)
3433.	SPGF4 (SYCP3)
3434.	SPGF5 (AURKC)
3435.	SPGF6 (SPATA16)
3436.	SPGF7 (CATSPER1)
3437.	SPGF8 (NR5A1)
3438.	SPGF9 (DPY19L2)
3439.	Spherocytosis type 1 (ANK1)
3440.	Spherocytosis type 2 (SPTB)
3441.	Spherocytosis type 3 (SPTA1)
3442.	Spherocytosis type 5 (EPB42)
3443.	Spheroid body myopathy (MYOT)
3444.	Spiegler-Brooke syndrome (CYLD)
3445.	Spina bifida folate sensitive (MTRR)
3446.	Spinal and bulbar muscular atrophy X-linked (AR)
3447.	Spinal muscular atrophy (SMA), NAIP related (NAIP)
3448.	Spinal muscular atrophy distal, autosomal recessive type 4 (PLEKHG5)
3449.	Spinal muscular atrophy type 1 (SMN1)
3450.	Spinal muscular atrophy type 2 (SMN1)
3451.	Spinal muscular atrophy type 3 (SMN1)
3452.	Spinal muscular atrophy type 3, modifier of (SMN2)
3453.	Spinal muscular atrophy type 4 (SMN1)
3454.	Spinal muscular atrophy type 5 (DNAJB2)

3455.	Spinal muscular atrophy with progressive myoclonic epilepsy (ASAH1)
3456.	Spinal muscular atrophy, distal, X-linked (ATP7A)
3457.	Spinal muscular atrophy, lower extremity, autosomal dominant, type 2 (BICD2)
3458.	Spinal muscular atrophy, lower extremity-predominant type 1, autosomal dominant (NGS Sequencing: DYNC1H1)
3459.	Spinocerebellar ataxia type 1, autosomal dominant (ATXN1)
3460.	Spinocerebellar ataxia type 1, X-linked (ATP2B3)
3461.	Spinocerebellar ataxia type 10, autosomal dominant (ATXN10)
3462.	Spinocerebellar ataxia type 10, autosomal recessive (ANO10)
3463.	Spinocerebellar ataxia type 11, autosomal dominant (TTBK2)
3464.	Spinocerebellar ataxia type 12, autosomal dominant (PPP2R2B)
3465.	Spinocerebellar ataxia type 12, autosomal recessive (WWOX)
3466.	Spinocerebellar ataxia type 13, autosomal dominant (KCNC3)
3467.	Spinocerebellar ataxia type 13, autosomal recessive (GRM1)
3468.	Spinocerebellar ataxia type 14, autosomal dominant (PRKCG)
3469.	Spinocerebellar ataxia type 15 (NGS Sequencing: ITPR1)
3470.	Spinocerebellar ataxia type 17, autosomal dominant (TBP)
3471.	Spinocerebellar ataxia type 17, autosomal recessive (CWF19L1)
3472.	Spinocerebellar ataxia type 18, autosomal dominant (IFRD1)
3473.	Spinocerebellar ataxia type 18, autosomal recessive (GRID2)
3474.	Spinocerebellar ataxia type 2, autosomal dominant (ATXN2)
3475.	Spinocerebellar ataxia type 21, autosomal dominant (TMEM240)
3476.	Spinocerebellar ataxia type 22, autosomal dominant (KCND3)
3477.	Spinocerebellar ataxia type 23, autosomal dominant (PDYN)
3478.	Spinocerebellar ataxia type 26, autosomal dominant (EEF2)
3479.	Spinocerebellar ataxia type 27, autosomal dominant (FGF14)
3480.	Spinocerebellar ataxia type 28, autosomal dominant (AFG3L2)
3481.	Spinocerebellar ataxia type 29, congenital nonprogressive (NGS Sequencing: ITPR1)
3482.	Spinocerebellar ataxia type 3, autosomal dominant (ATXN3)
3483.	Spinocerebellar ataxia type 31, autosomal dominant (BEAN1)
3484.	Spinocerebellar ataxia type 35, autosomal dominant (TGM6)
3485.	Spinocerebellar ataxia type 36, autosomal dominant (NOP56)
3486.	Spinocerebellar ataxia type 4, autosomal dominant (PLEKHG4)

3487.	Spinocerebellar ataxia type 5, autosomal dominant (SPTBN2)
3488.	Spinocerebellar ataxia type 6, autosomal dominant (CACNA1A)
3489.	Spinocerebellar ataxia type 7, autosomal dominant (ATXN7)
3490.	Spinocerebellar ataxia type 7, autosomal recessive (TPP1)
3491.	Spinocerebellar ataxia type 8, autosomal dominant (ATXN8OS)
3492.	Spinocerebellar ataxia type 8, autosomal recessive (NGS Sequencing: SYNE1)
3493.	Spinocerebellar ataxia type 9, autosomal rececssive (COQ8A)
3494.	Spinocerebellar ataxia with axonal neuropathy, autosomal recessive (TDP1)
3495.	Spinocerebellar ataxia, infantile-onset (TWNK)
3496.	Split-hand/foot malformation type 1 with sensorineural hearing loss (DLX5)
3497.	Split-hand/foot malformation type 6 (WNT10B)
3498.	Spondylo-megaepiphyseal-metaphyseal dysplasia (NKX3-2)
3499.	Spondylocarpotarsal synostosis syndrome (FLNB)
3500.	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like (SLC39A13)
3501.	Spondylocostal dysostosis type 5 (TBX6)
3502.	Spondylocostal dysostosis, autosomal recessive type 1 (DLL3)
3503.	Spondylocostal dysostosis, autosomal recessive type 2 (MESP2)
3504.	Spondylocostal dysostosis, autosomal recessive type 3 (LFNG)
3505.	Spondyloenchondrodysplasia with immune dysregulation (ACP5)
3506.	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures (B3GALT6)
3507.	Spondyloepimetaphyseal dysplasia, MATN3 related (MATN3)
3508.	Spondyloepiphyseal dysplasia with congenital joint dislocations (CHST3)
3509.	Spondylometaepiphyseal dysplasia, short limb-hand type (DDR2)
3510.	Spondyloperipheral dysplasia (COL2A1)
3511.	Squamous cell carcinoma, burn scar-related, somatic (FAS)
3512.	Stargardt Disease type 1 (ABCA4)
3513.	Stargardt Disease type 1 (CNGB3)
3514.	Stargardt Disease type 3 (ELOVL4)
3515.	Stargardt Disease type 4 (PROM1)
3516.	Steatocystoma multiplex (KRT17)
3517.	Stickler syndrome type 1 (COL2A1)
3518.	Stickler syndrome type 1, nonsyndromic ocular (COL2A1)

3519.	Stickler syndrome type 2 (NGS Sequencing: COL11A1)
3520.	Stickler syndrome type 3 (COL11A2)
3521.	Stickler syndrome type 5 (COL9A2)
3522.	Stickler syndrome, autosomal recessive (COL9A1)
3523.	Stiff skin syndrome (NGS Sequencing: FBN1)
3524.	Stocco dos Santos X-linked mental retardation syndrome (SHROOM4)
3525.	Stormorken syndrome (STIM1)
3526.	Striatal degeneration (PDE8B)
3527.	Stuve-Wiedemann syndrome (LIFR)
3528.	Succinic semialdehyde dehydrogenase deficiency (ALDH5A1)
3529.	Succinyl CoA:3-oxoacid CoA transferase deficiency (OXCT1)
3530.	Sucrase-isomaltase deficiency (SI)
3531.	Sudden infant death syndrome, susceptibility to (SCN5A)
3532.	Sudden infant death with dysgenesis of the testes syndrome (TSPYL1)
3533.	Sulfatase deficiency (SUMF1)
3534.	Sulfite oxidase deficiency (SUOX)
3535.	Supravalvar aortic stenosis (ELN)
3536.	Surfactant metabolism dysfunction (SFTPD)
3537.	Surfactant metabolism dysfunction type 1 (SFTPB)
3538.	Surfactant metabolism dysfunction type 2 (SFTPC)
3539.	Surfactant metabolism dysfunction type 3 (ABCA3)
3540.	Surfactant metabolism dysfunction type 4 (CSF2RA)
3541.	Surfactant metabolism dysfunction type 5 (CSF2RB)
3542.	Sveinsson choreoretinal atrophy (TEAD1)
3543.	Syndactyly type 1 (HOXD13)
3544.	Syndactyly type 5 (HOXD13)
3545.	Syndactyly, mesoaxial synostotic, with phalangeal reduction (BHLHA9)
3546.	Systemic lupus erythematosus (DNASE1)
3547.	Systemic lupus erythematosus type 16 (DNASE1L3)
3548.	Systemic lupus erythematosus, susceptibility to (ITGAM)
3549.	T-cell immunodeficiency, congenital alopecia, and nail dystrophy (FOXN1)
3550.	TANC2 related brain disorders (TANC2)

3551.	Tangier disease (ABCA1)
3552.	Tay-Sachs disease (HEXA)
3553.	Tay-Sachs disease AB variant (GM2A)
3554.	Telangiectasia hereditary hemorrhagic type 5 (GDF2)
3555.	Telangiectasia, hereditary hemorrhagic, of Rendu, Osler and Weber type 1 (ENG)
3556.	Telangiectasia, hereditary hemorrhagic, type 2 (ACVRL1)
3557.	Temple syndrome (maternal UPD chr. 14)
3558.	Temple-Baraitser syndrome (KCNH1)
3559.	Temtamy syndrome (C12orf57)
3560.	Terminal osseous dysplasis (FLNA)
3561.	Testicular anomalies with or without congenital heart disease (GATA4)
3562.	Testicular tumor, somatic (STK11)
3563.	Tetraamelia, autosomal recessive (WNT3)
3564.	Tetralogy of Fallot (ZFPM2)
3565.	Tetralogy of Fallot (ALDH1A2)
3566.	Tetralogy of Fallot (GATA6)
3567.	Tetralogy of Fallot (GATA4)
3568.	Thalassemia, alpha (HBA1)
3569.	Thalassemia, alpha (HBA2)
3570.	Thalassemia, delta (HBD)
3571.	Thiamine metabolism dysfunction syndrome 4 progressive polyneuropathy type (SLC25A19)
3572.	Thiamine metabolism dysfunction syndrome type 5 (TPK1)
3573.	Thiamine-responsive megaloblastic anemia syndrome (SLC19A2)
3574.	Thoracic aortic aneurysm dissection (SMAD2)
3575.	Three M syndrome type 1 (CUL7)
3576.	Three M syndrome type 2 (OBSL1)
3577.	Three M syndrome type 3 (CCDC8)
3578.	Thrombocythemia type 3, somatic (JAK2)
3579.	Thrombocytopenia congenital amegakaryocytic (MPL)
3580.	Thrombocytopenia type 2 (ANKRD26)
3581.	Thrombocytopenia type 2 (MASTL)
3582.	Thrombocytopenia type 4 (CYCS)

3583.	Thrombocytopenia type 5 (ETV6)
3584.	Thrombocytopenia with beta thalassemia X-linked (GATA1)
3585.	Thrombocytopenia, neonatal alloimmune (ITGB3)
3586.	Thrombocytopenia, neonatal alloimmune (ITGA2B)
3587.	Thrombocytopenia, X-linked (GATA1)
3588.	Thrombocytopenia, X-linked, intermittent (WAS)
3589.	Thrombocytosis, familial, JAK2 related (JAK2)
3590.	Thrombophilia due to thrombin defect (F2)
3591.	Thrombophilia, X-linked, due to factor IX defect (F9)
3592.	Thrombotic thrombocytopenic purpura (ADAMTS13)
3593.	Thromboxane synthase deficiency (TBXAS1)
3594.	Thromocytopenia-Absent-Radius-Syndrome (RBM8A)
3595.	Thyroid adenoma, hyperfunctioning, somatic (TSHR)
3596.	Thyroid cancer type 2, nonmedullary, susceptibility to (SRGAP1)
3597.	Thyroid carcinoma with thyrotoxicosis (TSHR)
3598.	Thyroid carcinoma, follicular, HRAS related, somatic (HRAS)
3599.	Thyroid carcinoma, follicular, somatic (NRAS)
3600.	Thyroid dyshormonogenesis type 1 (SLC5A5)
3601.	Thyroid dyshormonogenesis type 2A (TPO)
3602.	Thyroid dyshormonogenesis type 3 (TG)
3603.	Thyroid dyshormonogenesis type 4 (IYD)
3604.	Thyroid dyshormonogenesis type 5 (DUOXA2)
3605.	Thyroid dyshormonogenesis type 6 (DUOX1)
3606.	Thyroid dyshormonogenesis type 6 (DUOX2)
3607.	Thyroid hormone metabolism abnormal (SECISBP2)
3608.	Thyroid hormone resistance (THRB)
3609.	Thyrotoxic periodic paralysis type 1 (CACNA1S)
3610.	Thyrotoxic periodic paralysis type 2 (KCNJ18)
3611.	Tibial muscular dystrophy, tardive (NGS Sequencing: TTN)
3612.	Tietz albinism-deafness syndrome (MITF)
3613.	TJP1 deficiency (TJP1)
3614.	Toe syndactyly, telecanthus, and anogenital and renal malformations (FAM58A)

3615.	Tooth agenesis, selective type 1 (MSX1)
3616.	Tooth agenesis, selective type 3 (PAX9)
3617.	Tourette syndrome (SLITRK1)
3618.	Townes-Brocks syndrome (SALL1)
3619.	TPMT deficiency (TPMT)
3620.	Transaldolase deficiency (TALDO1)
3621.	Transcobalamin II deficiency (TCN2)
3622.	Transposition of great arteries, dextro-looped 3 (GDF1)
3623.	Transposition of the great arteries, dextro-looped 1 (MED13L)
3624.	Treacher Collins syndrome type 1 (TCOF1)
3625.	Treacher Collins syndrome type 2 (POLR1D)
3626.	Treacher Collins syndrome type 3 (POLR1C)
3627.	Tremor essential type 4 (FUS)
3628.	Trichodontoosseous syndrome (DLX3)
3629.	Trichoepithelioma, multiple familial, type 1 (CYLD)
3630.	Trichohepatoenteric syndrome type 1 (TTC37)
3631.	Trichohepatoenteric syndrome type 2 (SKIV2L)
3632.	Trichorhinophalangeal syndrome type 1 (TRPS1)
3633.	Trichothiodystrophy (ERCC3)
3634.	Trichothiodystrophy (ERCC2)
3635.	Trichothiodystrophy (GTF2H5)
3636.	Trichothiodystrophy, nonphotosensitive type 1 (MPLKIP)
3637.	Trifunctional protein deficiency (HADHA)
3638.	Trifunctional protein deficiency (HADHB)
3639.	Trigonocephaly type 1 (FGFR1)
3640.	Trimethylaminuria (FMO3)
3641.	Triosephosphate isomerase deficiency (TPI1)
3642.	Tuberous sclerosis (TSC1)
3643.	Tuberous sclerosis type 2 (TSC2)
3644.	Tuftelin deficiency (TUFT1)
3645.	Tumor predisposition syndrome (BAP1)
3646.	Tumor predisposition syndrome, ARL11 related (ARL11)

3647.	Tylosis with esophageal cancer (RHBDF2)
3648.	Tyrosine kinase 2 deficiency (TYK2)
3649.	Tyrosinemia type 1 (FAH)
3650.	Tyrosinemia type 1B (GSTZ1)
3651.	Tyrosinemia type 2 (TAT)
3652.	Tyrosinemia type 3 (HPD)
3653.	Ullrich congenital muscular dystrophy (COL6A2)
3654.	Ullrich congenital muscular dystrophy (COL6A1)
3655.	Ullrich congenital muscular dystrophy type 1 (COL6A3)
3656.	Ullrich congenital muscular dystrophy type 2 (NGS Sequencing: COL12A1)
3657.	Ulna and fibula, absence of, with severe limb deficiency (WNT7A)
3658.	Ulnar-Mammary syndrome (TBX3)
3659.	Unverricht-Lundborg disease (CSTB)
3660.	Urbach-Wiethe disease (ECM1)
3661.	Urocanase deficiency (UROC1)
3662.	Urofacial syndrome (LRIG2)
3663.	Usher syndrome type 1D/F (PCDH15)
3664.	Usher syndrome type 1G (USH1G)
3665.	Usher syndrome type 1J (CIB2)
3666.	Usher syndrome type 2C (NGS Sequencing: ADGRV1)
3667.	Usher syndrome type 2C (PDZD7)
3668.	Usher syndrome type 3A (CLRN1)
3669.	Usher syndrome, type 1D (NGS Sequencing: CDH23)
3670.	UV-sensitive syndrome type 1 (ERCC6)
3671.	UV-sensitive syndrome type 3 (UVSSA)
3672.	Uveal melanoma, GNAQ related, somatic (GNAQ)
3673.	Van Buchem disease (SOST)
3674.	Van den Ende-Gupta syndrome (SCARF2)
3675.	van der Woude syndrome type 1 (IRF6)
3676.	van der Woude syndrome type 2 (GRHL3)
3677.	Van Maldergem syndrome type 2 (FAT4)
3678.	Various cancers, DICER1 related, somatic (DICER1)

3679.	Vascular system defects due to CALCRL deficiency (CALCRL)
3680.	Vascular system defects due to GNA13 deficiency (GNA13)
3681.	Vasculopathy, infantile-onset, TMEM173/STING related (TMEM173)
3682.	Vater association (HOXD13)
3683.	Ventricular fibrillation, paroxysmal familial type 1 (SCN5A)
3684.	Ventricular septal defect type 1 (GATA4)
3685.	Ventricular septal defect type 2 (CITED2)
3686.	Ventricular tachycardia, catecholaminergic polymorphic type 1 (NGS Sequencing: RYR2)
3687.	Ventricular tachycardia, catecholaminergic polymorphic type 2 (CASQ2)
3688.	Ventricular tachycardia, catecholaminergic polymorphic type 4 (CALM1)
3689.	Ventricular tachycardia, catecholaminergic polymorphic type 5 (TRDN)
3690.	Ventriculomegaly with cystic kidney disease (CRB2)
3691.	Vesicoureteral reflux type 2 (ROBO2)
3692.	Vesicoureteral reflux type 3 (SOX17)
3693.	Vici syndrome (EPG5)
3694.	Visceral myopathy (ACTG2)
3695.	Vitamin E familial deficiency (TTPA)
3696.	Vitamin K-dependent clotting factors combined deficiency type 1 (GGCX)
3697.	Vitiligo-associated multiple autoimmune disease (NLRP1)
3698.	Vitreoretinochoroidopathy (BEST1)
3699.	Vohwinkel syndrome with ichthyosis (LOR)
3700.	von Hippel-Lindau syndrome (VHL)
3701.	von Willebrand disease (VWF)
3702.	von Willebrand disease platelet type (GP1BA)
3703.	Von-Gierke disease (G6PC)
3704.	Waardenburg syndrome type 1 (PAX3)
3705.	Waardenburg syndrome type 2D (SNAI2)
3706.	Waardenburg syndrome type 2E (SOX10)
3707.	Waardenburg syndrome type 4C (SOX10)
3708.	Waardenburg syndrome/albinism (MITF)
3709.	Waardenburg syndrome/albinism (TYR)
3710.	Waardenburg syndrome/Hirschsprung disease (EDNRB)

3711.	Wagner syndrome (VCAN)
3712.	Walker-Warburg syndrome (ISPD)
3713.	Walker-Warburg syndrome (FKTN)
3714.	Walker-Warburg syndrome or muscle-eye-brain disease, FKRP related (FKRP)
3715.	Warburg micro syndrome 3 (RAB18)
3716.	Warburg micro syndrome type 1 (RAB3GAP1)
3717.	Warburg micro syndrome type 2 (RAB3GAP2)
3718.	Warsaw breakage syndrome (DDX11)
3719.	WDR27-related brain disorders (WDR27)
3720.	Weaver syndrome (EZH2)
3721.	Webb-Dattani syndrome (ARNT2)
3722.	Weill-Marchesani syndrome - AR (ADAMTS10)
3723.	Weill-Marchesani syndrome type 3 (LTBP2)
3724.	Weill-Marchesani syndrome, dominant type 2 (NGS Sequencing: FBN1)
3725.	Werner syndrome (WRN)
3726.	WHIM syndrome (CXCR4)
3727.	Wieacker-Wolff syndrome (ZC4H2)
3728.	Wiedemann-Steiner syndrome (KMT2A)
3729.	Williams-Beuren syndrome (chr. 7q11.23)
3730.	Wilms tumor type 1, familial (WT1)
3731.	Wilms tumor, familial, due to BRCA2 mutation (BRCA2)
3732.	Wilms tumor, IGF2 related, somatic (IGF2)
3733.	Wilson disease (ATP7B)
3734.	Winchester Syndrome (MMP14)
3735.	Witkop syndrome (MSX1)
3736.	Wolcott-Rallison syndrome (EIF2AK3)
3737.	Wolff -Parkinson-White syndrome (PRKAG2)
3738.	Wolfram syndrome type 1 (WFS1)
3739.	Wolfram syndrome type 2 (CISD2)
3740.	Wolfram-like syndrome, autosomal dominant (WFS1)
3741.	Wolman disease (LIPA)
3742.	Wrinkly skin syndrome (ATP6V0A2)

3743.	Xanthinuria type 1 (XDH)
3744.	Xeroderma pigmentosum, group A (XPA)
3745.	Xeroderma pigmentosum, group C (XPC)
3746.	Xeroderma pigmentosum, group D (ERCC2)
3747.	Xeroderma pigmentosum, group E, DDB-negative subtype (DDB2)
3748.	Xeroderma pigmentosum, group F (ERCC4)
3749.	Xeroderma pigmentosum, group G (ERCC5)
3750.	Xeroderma pigmentosum, variant type (POLH)
3751.	XFE progeroid syndrome (ERCC4)
3752.	Zellweger syndrome (PEX26)
3753.	Zellweger syndrome (PEX1)
3754.	Zellweger syndrome (PEX2)
3755.	Zellweger syndrome (PEX6)
3756.	Zellweger syndrome (PEX10)
3757.	Zellweger syndrome (PEX12)
3758.	Zellweger syndrome (PEX13)
3759.	Zellweger syndrome (PEX14)
3760.	Zellweger syndrome (PEX16)
3761.	Zellweger syndrome (PEX19)
3762.	ZIC5 related brain disorders (ZIC5)

GENERAL INFORMATION

- PRICES
- TURN AROUND TIME
- SAMPLING PROCEDURE
- REPORTING PROCEDURE AND SAMPLE REPORTS
- FLOW OF INFORMATION

Prices

Prices to be provided upon request.

Turn Around Time

The turnaround time of the tests varies from minimum 2 weeks to maximum 5 weeks depending upon the test.

Sample Collection

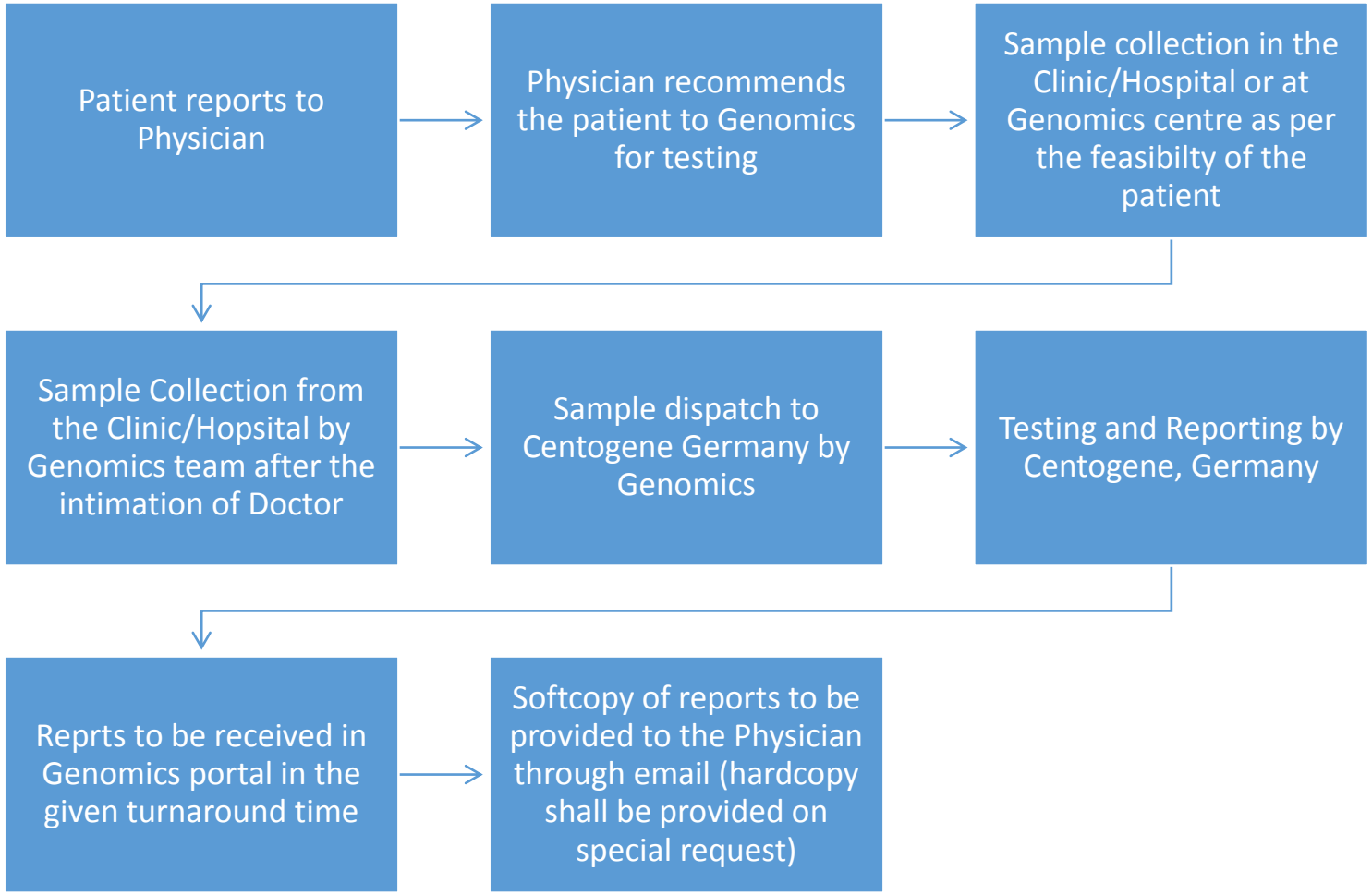
Except for NIPT, all other tests are dried blood samples. Blood is to be drawn in purple tube(EDTA) before putting 4 drops on each circle of the provided **centocard**. The sample is allowed to dry for 3 hours.

NOTE: the forms attached and provided along with the Centocard must be thoroughly completed and all the clinical information must be provided for the proper diagnosis.

Reporting

The reports shall be received in Genomics Central Portal within the turnaround time and will be sent to the Physician through an email. Hardcopy of the reports can also be provided to the physician on their request.

Flow of Information



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Decoding the Human Genome

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