

## *Supplementary Information*

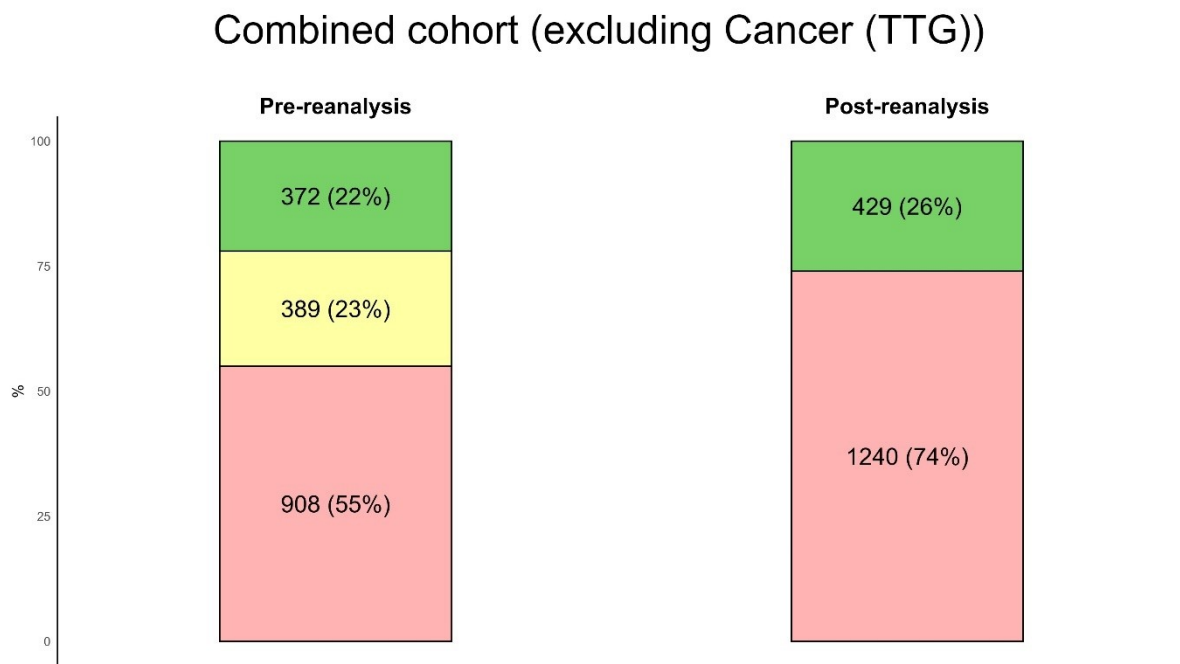
Figure S1: Diagnostic yield for cohort pre- and post-reanalysis, excluding cancer TTG.

Figure S2: Variant evaluation workflow

Table S1: *In silico* gene panel creation strategy per patient group

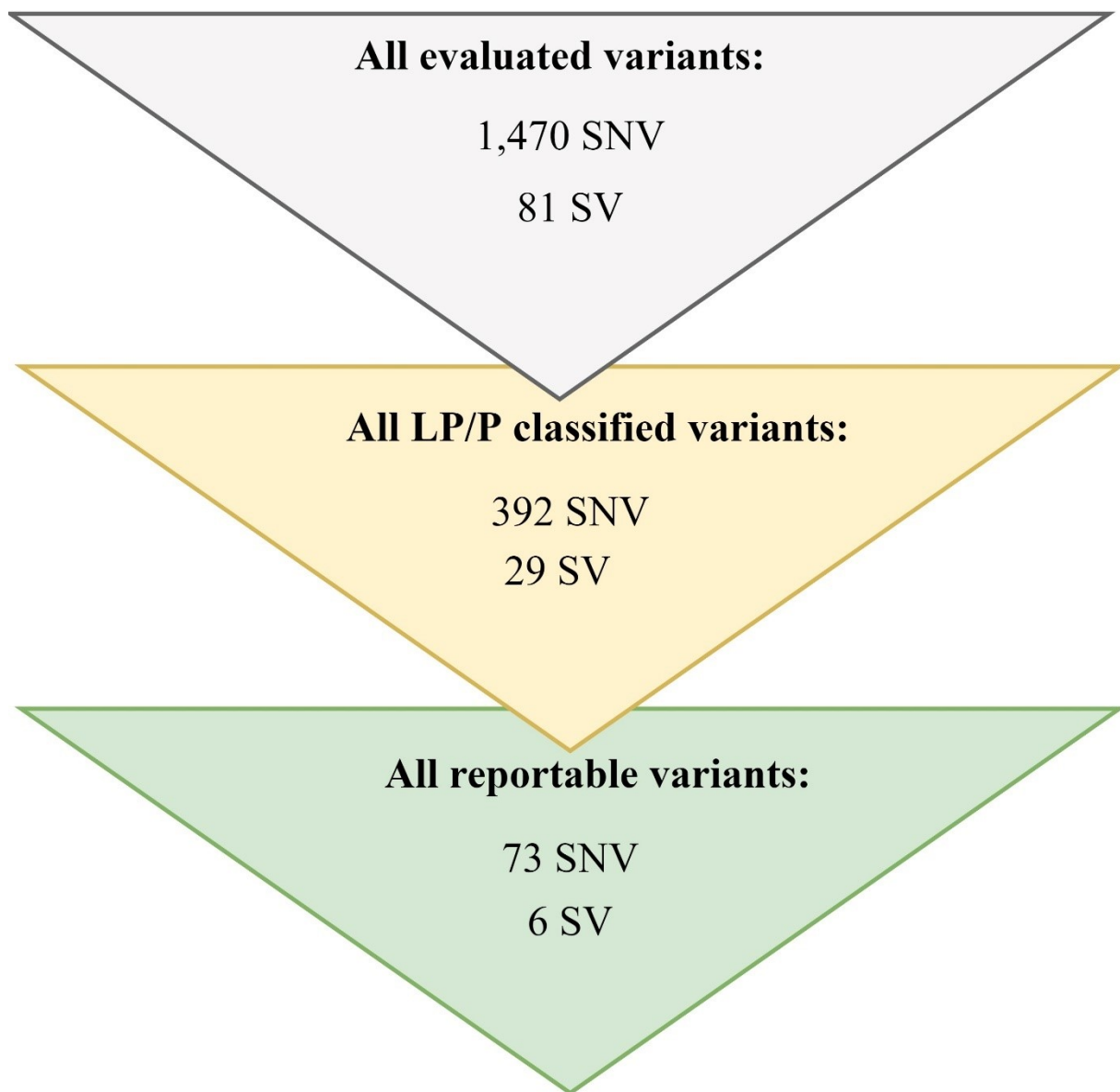
Table S2: List of Genomics England PanelApp *in silico* gene panels applied during reanalysis

Table S3: Genes contained in the applied reanalysis *in silico* gene panels



**Figure S1: Diagnostic yield for cohort pre- and post-reanalysis, excluding cancer TTG.**

Exclusion of the cancer targeted treatment patient group from the combined cohort results in an overall increase in the diagnostic yield from 22% pre-reanalysis to 26% post-reanalysis.



**Figure S2: Variant evaluation workflow**

A total of 1,470 SNVs and 81 SVs were manually curated. Of these, 421 variants were classified as LP/P. 79 variants were considered clinically reportable.

| Patient group                | Strategy applied                                                                                                                                                                                                                                                                                                                                                                     |
|------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Cancer                       | Genes were sourced from 39 Genomics England PanelApp cancer-related gene panels. Duplicate genes were removed from the final list and a gene panel of 180 unique genes was created. This gene panel was applied to all patients in the cancer patient group.                                                                                                                         |
| Endocrine disease            | Genes were sourced from 12 Genomics England PanelApp endocrine disease-related gene panels. Duplicate genes were removed from the final list and a gene panel of 176 unique genes was created. This gene panel was applied to all patients in the endocrine disease patient group.                                                                                                   |
| Hematological disease        | Genes were sourced from 6 Genomics England PanelApp hematological disease-related gene panels. One or more of these gene panels were applied to patients in the hematological disease patient group, depending on the specific sub-phenotype of the patient. For the subgroups Anemia and Platelet and bleeding disorders, two gene panels were used to create the final gene panel. |
| Neurodevelopmental Disorders | Genes were sourced from 3 Genomics England PanelApp neurodevelopment disorders-related gene panels. Duplicate genes were removed from the final list and a gene panel of 2259 unique genes was created. This gene panel was applied to all patients in the neurodevelopmental disorders patient group.                                                                               |

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| Primary immunodeficiency | Genes were sourced from 3 Genomics England PanelApp primary immunodeficiency-related gene panels. Duplicate genes were removed from the final list and a gene panel of 362 unique genes was created. This gene panel was applied to all patients in the primary immunodeficiency patient group.                                                         |
| Miscellaneous            | For most patients in the miscellaneous patient group, with a phenotype of cholestasis or pancreatitis, a relevant Genomics England PanelApp gene panel was applied (listed in Table S2/S3). For the minority of patients with other phenotypes a relevant Genomics England PanelApp gene panel was applied (specific panels not listed in Table S2/S3). |

**Table S1: *In silico* gene panel creation strategy per patient group**

*In silico* gene panels were sourced from Genomics England PanelApp and gene panels appropriate for the specific patient groups were selected. For all patient groups, except hematological disease and miscellaneous, a single combined final panel to be used for the WGS reanalysis was created by merging individual gene panels and removing duplicate genes.

| Patient group | Patient subgroup | Genomics England PanelApp gene panel name                                | Version |
|---------------|------------------|--------------------------------------------------------------------------|---------|
| Cancer        | -                | Adult solid tumours cancer susceptibility                                | v2.25   |
| Cancer        | -                | Adult solid tumours for rare disease                                     | v1.37   |
| Cancer        | -                | Bladder cancer pertinent cancer susceptibility                           | v1.1    |
| Cancer        | -                | Brain cancer pertinent cancer susceptibility                             | v1.2    |
| Cancer        | -                | Breast cancer pertinent cancer susceptibility                            | v2.5    |
| Cancer        | -                | Childhood solid tumours                                                  | v4.7    |
| Cancer        | -                | Childhood solid tumours cancer susceptibility                            | v1.25   |
| Cancer        | -                | Colorectal cancer pertinent cancer susceptibility                        | v1.0    |
| Cancer        | -                | Endometrial cancer pertinent cancer susceptibility                       | v1.0    |
| Cancer        | -                | Endocrine neoplasia                                                      | v3.1    |
| Cancer        | -                | Familial prostate cancer                                                 | v1.3    |
| Cancer        | -                | Familial breast cancer                                                   | v1.20   |
| Cancer        | -                | Familial melanoma                                                        | v2.1    |
| Cancer        | -                | Familial rhabdomyosarcoma                                                | v1.5    |
| Cancer        | -                | Familial Tumours Syndromes of the central & peripheral Nervous system    | v1.14   |
| Cancer        | -                | Genodermatoses with malignancies                                         | v1.10   |
| Cancer        | -                | GI tract tumours                                                         | v1.22   |
| Cancer        | -                | Haematological malignancies cancer susceptibility                        | v4.2    |
| Cancer        | -                | Head and neck cancer pertinent cancer susceptibility                     | v1.0    |
| Cancer        | -                | Inherited breast cancer and ovarian cancer                               | v2.7    |
| Cancer        | -                | Inherited ovarian cancer (without breast cancer)                         | v4.1    |
| Cancer        | -                | Inherited pancreatic cancer                                              | v2.1    |
| Cancer        | -                | Inherited polyposis and early onset colorectal cancer - germline testing | v2.6    |
| Cancer        | -                | Inherited non-medullary thyroid cancer                                   | v1.5    |
| Cancer        | -                | Inherited pheochromocytoma and paraganglioma                             | v1.11   |
| Cancer        | -                | Inherited prostate cancer                                                | v1.2    |
| Cancer        | -                | Inherited renal cancer                                                   | v1.24   |
| Cancer        | -                | Melanoma pertinent cancer susceptibility                                 | v1.0    |
| Cancer        | -                | Multiple endocrine tumours                                               | v1.14   |

|               |                                 |                                                               |        |
|---------------|---------------------------------|---------------------------------------------------------------|--------|
| Cancer        | -                               | Neuroendocrine cancer pertinent cancer susceptibility         | v1.4   |
| Cancer        | -                               | Neurofibromatosis Type 1                                      | v1.32  |
| Cancer        | -                               | Ovarian cancer pertinent cancer susceptibility                | v2.1   |
| Cancer        | -                               | Prostate cancer pertinent cancer susceptibility               | v1.2   |
| Cancer        | -                               | Parathyroid Cancer                                            | v1.4   |
| Cancer        | -                               | Renal cancer pertinent cancer susceptibility                  | v1.2   |
| Cancer        | -                               | Sarcoma cancer susceptibility                                 | v1.21  |
| Cancer        | -                               | Sarcoma susceptibility                                        | v1.81  |
| Cancer        | -                               | Thyroid cancer pertinent cancer susceptibility                | v1.3   |
| Cancer        | -                               | Upper gastrointestinal cancer pertinent cancer susceptibility | v1.0   |
| Endocrine     | -                               | Congenital hypothyroidism                                     | v2.13  |
| Endocrine     | -                               | Endocrine neoplasia                                           | v3.1   |
| Endocrine     | -                               | Extreme early-onset hypertension                              | v1.21  |
| Endocrine     | -                               | Familial hyperparathyroidism or hypocalciuric hypercalcaemia  | v3.3   |
| Endocrine     | -                               | Familial hypoparathyroidism                                   | v2.10  |
| Endocrine     | -                               | Hypophosphataemia or rickets                                  | v3.1   |
| Endocrine     | -                               | Hypogonadotropic hypogonadism                                 | v3.4   |
| Endocrine     | -                               | Hyperthyroidism                                               | v3.1   |
| Endocrine     | -                               | Inherited pheochromocytoma and paraganglioma                  | v1.11  |
| Endocrine     | -                               | Multiple endocrine tumours                                    | v1.14  |
| Endocrine     | -                               | Osteogenesis imperfecta                                       | v4.5   |
| Endocrine     | -                               | Renal tubulopathies                                           | v4.17  |
| Erythrosis    | -                               | Hereditary Erythrocytosis                                     | v2.1   |
| Hematological | Anemia                          | Cytopenias and congenital anaemias                            | v1.111 |
| Hematological | Anemia                          | Rare anaemia                                                  | v3.8   |
| Hematological | Erythrocytosis                  | Hereditary Erythrocytosis                                     | v2.6   |
| Hematological | Hemochromatosis                 | Iron metabolism disorders - NOT common HFE mutations          | v2.1   |
| Hematological | Platelet and bleeding disorders | Bleeding and platelet disorders                               | v3.1   |

|                              |                                 |                                                                  |        |
|------------------------------|---------------------------------|------------------------------------------------------------------|--------|
| Hematological                | Platelet and bleeding disorders | Inherited bleeding disorders                                     | v1.178 |
| Neurodevelopmental Disorders | -                               | Autism                                                           | v0.36  |
| Neurodevelopmental Disorders | -                               | DDG2P                                                            | v3.1   |
| Neurodevelopmental Disorders | -                               | Intellectual disability                                          | v6.68  |
| Primary immunodeficiency     | -                               | Primary immunodeficiency or monogenic inflammatory bowel disease | v5.3   |
| Primary immunodeficiency     | -                               | Autoinflammatory disorders                                       | v2.3   |
| Primary immunodeficiency     | -                               | Infantile enterocolitis & monogenic inflammatory bowel disease   | v1.44  |
| Miscellaneous                | Cholestasis                     | Cholestasis                                                      | v3.4   |
| Miscellaneous                | Pancreatitis                    | Pancreatitis                                                     | v3.4   |

**Table S2: List of Genomics England PanelApp *in silico* gene panels applied during reanalysis**

*In silico* gene panels were sourced from Genomics England PanelApp using the latest available version at the time of analysis. The number of PanelApp gene panels used to create the final *in silico* gene panel per patient group ranged from 1 to 39.

| <i>In silico</i> gene panel | Genes                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |
|-----------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Cancer                      | APC, ACD, AIP, ALK, ANKRD26, ATM, BAP1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CHEK2, CTC1, CTR9, DDB2, DDX41, DICER1, DIS3L2, DKC1, DNAJC21, DOCK8, ELANE, ELP1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6L2, ETV6, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAS, FH, FLCN, GATA1, GATA2, GBA, GNAS, GPC3, GPR161, HAX1, HRAS, IKZF1, ITK, KIT, KITLG, KRAS, LIG4, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAX, MBD4, MEN1, MET, MLH1, MSH2, MSH6, MTAP, MUTYH, NAF1, NBN, NF1, NF2, NHP2, NOP10, NRAS, NSD1, NTHL1, PALB2, PARN, PAX5, PDGFRA, PDGFRB, PHOX2B, PMS2, POLD1, POLE, POLH, POT1, PRF1, PRKAR1A, PTCH1, PTEN, PTPN11, RAD21, RAD51C, RAD51D, RAF1, RB1, RECQL4, REST, RET, RMRP, RNF43, RPL11, RPL15, RPL23, RPL26, RPL31, RPL35A, RPL36, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SH2D1A, SHOC2, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SOS1, SPRED1, SQSTM1, STAT3, STK11, STN1, SUFU, TERC, TERT, TINF2, TMEM127, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WAS, WRAP53, WRN, WT1, XPA, XPC, XRCC2.                                             |
| Endocrine disease           | AIP, AIRE, ALB, ALPL, ANOS1, AP2S1, AQP2, ATP1A1, ATP6V0A4, ATP6V1B1, AVPR2, B3GALT6, B4GALT7, BMP1, BSND, CA2, CASR, CDC73, CDKN1B, CHD7, CLCNKB, CLDN10, CLDN16, CLDN19, CNNM2, COL1A1, COL1A2, COPB2, CREB3L1, CRTAP, CTNS, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, CYP27B1, CYP2R1, DMP1, DUOX2, DUOX2A2, ENPP1, FAH, FAM20C, FGF23, FGF8, FGFR1, FH, FKBP10, FOXE1, FSHB, GATA3, GATM, GCM2, GLIS3, GNA11, GNAS, GNRH1, GNRHR, GORAB, HESX1, HNF1B, HNF4A, HSD11B2, IFITM5, IGSF1, IL17RD, IRS4, IYD, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KDELR2, KISS1R, KLB, KLHL3, LEP, LEPR, LHB, LHX3, LHX4, LRP5, MAGED2, MAX, MEN1, MESD, MTX2, NBAS, NDNF, NF1, NKX2-1, NOTCH2, NR3C1, NR3C2, OCRL, OTX2, P3H1, P4HB, PAX8, PHEX, PLOD2, PLS3, PLXNA3, POU1F1, PPIB, PRKAR1A, PROK2, PROKR2, PROP1, PTH, REN, RET, RRAGD, SARS2, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC24D, SEC61A1, SECISBP2, SEMA3F, SERPINF1, SERPINH1, SLC12A1, SLC12A3, SLC16A2, SLC22A12, SLC26A4, SLC26A7, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC4A1, SLC4A4, SLC5A2, SLC5A5, SOX10, SP7, SPARC, TAC3, TACR3, TAPT1, TBCE, TBL1X, TENT5A, TG, THRA, THRB, TMEM127, TMEM38B, TP53, TPO, TRHR, TRPM6, TRPV6, TSHB, TSHR, TTC21B, TTR, UMOD, UNC45A, VDR, VHL, VIPAS39, VPS33B, WDR11, WNK1, WNK4, WNT1. |

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| Anemia<br>(Hematological disease)                          | ABCB7, ABCG5, ABCG8, ADA2, AK1, ALAS2, ALDOA, AMN, ANK1, CBLIF, CD59, CDAN1, CUBN, CDIN1, CYB5R3, DHFR, EPB41, EPB42, G6PD, GATA1, GCLC, GLRX5, GPI, GSR, GSS, HBA1, HBA2, HBB, HBD, HBG1, HBG2, HEATR3, HK1, HSPA9, KCNN4, KIF23, KLF1, LARS2, LPIN2, MTR, MTRR, NT5C3A, PFKM, PIEZO1, PKLR, PUS1, RHAG, RPL11, RPL15, RPL31, RPL35A, RPL5, RPL9, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7, SBDS, SEC23B, SLC11A2, SLC19A2, SLC25A38, SLC2A1, SLC4A1, SPTA1, SPTB, TCN2, TF, TMPRSS6, TPI1, TRNT1, UMPS, VPS4A, XK, YARS2, ADAMTS13, BRCA2, BRIP1, CSF3R, CTC1, CXCR4, DKC1, DNAJC21, ELANE, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC3, GATA2, GFI1, HAX1, MPL, NBN, NHP2, PALB2, RMRP, RTEL1, SAMD9, SAMD9L, SLX4, TERT, UBE2T, UROS, VPS45, WAS, WIPF1. |
| Erythrocytosis<br>(Hematological disease)                  | BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, SLC30A10, VHL.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               |
| Hemochromatosis<br>(Hematological disease)                 | ABCB7, ALAS2, ATP7B, BMP6, CP, CYBRD1, FTL, GBA, GLRX5, HAMP, HFE, HJV, SLC11A2, SLC25A38, SLC40A1, TF, TFR2, TMPRSS6.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       |
| Platelet and bleeding disorders<br>(Hematological disease) | ABCG5, ABCG8, ACTB, ACTN1, ACVRL1, ADAMTS13, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S6, CDC42, CHST14, COL1A1, COL3A1, COL5A1, COL5A2, CYCS, DIAPH1, DTNBP1, ENG, ETV6, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FERMT3, FGA, FGB, FGG, FLI1, FLNA, FYB1, GATA1, GBA, GFI1B, GGCX, GNE, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRG, IKZF5, ITGA2B, ITGB3, KDSR, KLKB1, KNG1, LMAN1, LYST, MCFD2, MECOM, MPIG6B, MPL, MYH9, NBEA, NBEAL2, P2RY12, PLA2G4A, PLAT, PLAU, PLG, PROC, PROS1, PTGS1, PTPN11, RASGRP2, RBM8A, RNU4ATAC, RUNX1, SERPINC1, SERPIND1, SERPINE1, SERPINF2, SLC45A2, SLFN14, SMAD4, SRC, STIM1, STXBP2, TBXA2R, TBXAS1, THBD, THPO, TNXB, TPM4, TUBB1, VIPAS39, VKORC1, VPS33B, VWF, WAS.                                                      |
| Neurodevelopmental Disorders                               | ABAT, ABCA2, ABCB11, ABCB7, ABCC6, ABCC9, ABCD1, ABCD4, ABHD16A, ABHD5, ABL1, ACACA, ACAD9, ACADM, ACADS, ACADVL, ACAN, ACAT1, ACBD5, ACER3, ACO2, ACOX1, ACP5, ACSL4, ACTA1, ACTA2, ACTB, ACTG1, ACTL6A, ACTL6B, ACVR1, ACY1, ADA, ADAM22, ADAMTS18, ADAMTS9, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY5, ADD1, ADD3, ADGRG1, ADGRG6, ADK, ADNP, ADPRS, ADSL, AFF2, AFF3, AFF4, AGA, AGK, AGL, AGO1, AGO2, AGPS, AGTPBP1, AGXT, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIPL1, AIRE, AK2, AKR1D1, AKT1, AKT2, AKT3, ALDH18A1, ALDH1A2, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, ALPL, ALS2, ALX1, ALX3, ALX4, AMER1,                                                                                                                  |

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|  | <p> AMOTL1, AMPD2, AMT, ANAPC1, ANK2, ANK3, ANKH, ANKRD11, ANKRD17, ANKRD26, ANO5, ANTXR1, AP1G1, AP1S1, AP1S2, AP2M1, AP2S1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, AR, ARCN1, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGAP35, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARMC9, ARPC4, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASH1L, ASL, ASNS, ASPA, ASPH, ASPM, ASS1, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATG7, ATIC, ATL1, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP5F1A, ATP5F1D, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B1, ATP6V1B2, ATP6V1E1, ATP7A, ATP8A2, ATP8B1, ATP9A, ATR, ATRX, AUH, AUTS2, B3GALNT2, B3GALT6, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B9D1, B9D2, BANF1, BAP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCS1L, BFSP2, BGN, BHLHA9, BICD2, BICRA, BIN1, BLM, BLOC1S1, BLOC1S6, BLTP1, BMP2, BMP4, BMPER, BMPR1B, BNC2, BOLA3, BPNT2, BPTF, BRAF, BRAT1, BRCA1, BRCA2, BRD4, BRF1, BRIP1, BRPF1, BRSK2, BRWD3, BSCL2, BSND, BTB, BUB1, BUB1B, C12orf4, C12orf57, C1QBP, C2CD3, C2orf69, CA2, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1I, CACNA2D1, CAD, CAMK2A, CAMK2B, CAMK2G, CAMK4, CAMSAP1, CAMTA1, CANT1, CAPN15, CAPRIN1, CARS1, CARS2, CASK, CASP2, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC115, CCDC22, CCDC32, CCDC39, CCDC40, CCDC47, CCDC65, CCDC78, CCDC8, CCDC82, CCDC88C, CCND2, CCNO, CCNQ, CD151, CDC42, CDC45, CDC6, CDH1, CDH11, CDH2, CDH23, CDH3, CDK10, CDK13, CDK16, CDK19, CDK5RAP2, CDK8, CDKL5, CDKN1C, CDON, CDT1, CELF2, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP290, CEP41, CEP55, CEP57, CEP63, CEP83, CEP85L, CERT1, CFAP298, CFAP300, CFAP410, CFAP418, CFC1, CFL2, CHAMP1, CHD1, CHD2, CHD3, CHD4, CHD5, CHD7, CHD8, CHKA, CHKB, CHM, CHMP1A, CHRDL1, CHRNA1, CHRNA3, CHRNA4, CHRN1, CHRN2, CHRN3, CHST14, CHST3, CHSY1, CHUK, CIB2, CIC, CISD2, CIT, CKAP2L, CLCN3, CLCN4, CLCN6, CLCN7, CLCNKB, CLDN11, CLDN19, CLDN5, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, CNKSR2, CNNM2, CNOT1, CNOT2, CNOT3, CNOT9, CNPY3, CNTNAP1, CNTNAP2, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL13A1, COL18A1, COL1A1, COL25A1, COL27A1, COL2A1, COL4A1, COL4A2, COL4A3, COL4A4, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COPB2, COQ2, COQ4, COQ8A, COQ9, COX10, COX11, COX15, COX16, COX6B1, COX7B, CPAMD8, CPE, CPLANE1, CPLX1, CPS1, CPSF3, CRADD, CRB1, CRB2, CREBBP, CRELD1, CRPPA, CRX, CRYBA1, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYAA, CSDE1, CSF1R, CSNK1G1, CSNK2A1, CSNK2B, CSPP1, CSTA, CSTB, CTBP1, CTC1, CTCF, CTD1, CTNNA2, CTNNA1, CTNND1, CTNND2, CTNS, CTR9, CTSA, CTSD, CTSK, CTU2, CUL3, CUL4B, CUL7, CUX1, CUX2, CWC27, CWF19L1, CYB5R3, CYC1, CYFIP2, CYP1B1, CYP27A1, CYP2U1, D2HGDH, DAG1, DAGLA, DARS1, DARS2, DAW1, DBT, DCAF17, DCC, DCDC2, DCHS1, DCPS, DCX, DDB1, DDB2, DDC, DDHD1, DDHD2, DDOST, DDR2, DDX11, DDX23, DDX3X, DDX59, DDX6, DEAF1, DEGS1, DENND5A, DEPDC5, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHPS, DHTKD1, DHX30, DHX37, DHX9, DIAPH1, DIS3L2, DKC1, DLAT, DLD, DLG3, DLG4, DLL1, DLL3, DLL4, DMD, DMP1, DMPK, DMXL2, DNAH5, DNAH9, DNAJC12, DNAJC19, DNMI1, </p> |
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|  | <p> DNM1L, DNMT3A, DNMT3B, DNAAF11, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DOCK3, DOCK6, DOCK7, DOCK8, DOHH, DOLK, DPAGT1, DPF2, DPH1, DPH5, DPM1, DPM2, DPM3, DPYD, DPYS, DPYSL5, DRC1, DSCAM, DSG1, DSP, DSPP, DSTYK, DTYMK, DVL1, DVL3, DYM, DYNC1H1, DYNC1I2, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYRK1A, EARS2, EBF3, EBP, ECEL1, ECHS1, ECM1, EDA, EDAR, EDEM3, EDN1, EDNRA, EDNRB, EED, EEF1A2, EFEMP2, EFN1, EFTUD2, EGR2, EHMT1, EIF2AK2, EIF2AK3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EIF4A3, EIF5A, ELAC2, ELMO2, ELN, ELOVL4, ELP2, EMC1, EMC10, EMG1, EML1, EMX2, ENPP1, ENTPD1, EOGT, EP300, EPCAM, EPG5, EPRS1, ERBB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERF, ERI1, ERLIN2, ESAM, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EXOSC9, EXPH5, EXT1, EXT2, EXTL3, EYA1, EZH2, FAH, FAM111A, FAM126A, FAM149B1, FAM161A, FAM20A, FAM20C, FAM50A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAR1, FARS2, FARSA, FAT4, FBN1, FBN2, FBP1, FBRSL1, FBXL3, FBXL4, FBXO11, FBXO28, FBXO31, FBXW11, FBXW7, FCSK, FEZF1, FGD1, FGF10, FGF12, FGF13, FGF14, FGF3, FGF9, FGFR1, FGFR2, FGFR3, FH, FHL1, FIG4, FILIP1, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLT4, FLVCR1, FLVCR2, FMN2, FMR1, FN1, FOLR1, FOXC1, FOXC2, FOXE1, FOXE3, FOXF1, FOXG1, FOXJ1, FOXL2, FOXN1, FOXP1, FOXP2, FOXP3, FOXRED1, FRA10AC1, FRAS1, FREM1, FREM2, FRMD5, FRMPD4, FRRS1L, FTCD, FTL, FTSJ1, FUCA1, FUT8, FXR1, FYCO1, FZD5, FZD6, FZR1, GABBR1, GABBR2, GABRA1, GABRA2, GABRA5, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALE, GALK1, GALNS, GALNT2, GALT, GAMT, GAN, GAS2L2, GAS8, GATA2, GATA3, GATA4, GATA6, GATAD2B, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCSH, GDF1, GDF11, GDF5, GDF6, GDI1, GEMIN4, GEMIN5, GFAP, GFER, GFM1, GHR, GIGYF1, GJA1, GJA3, GJA8, GJB3, GJC2, GK, GLB1, GLDC, GLDN, GLE1, GLI2, GLI3, GLIS2, GLIS3, GLMN, GLRA1, GLRA2, GLRB, GLUD1, GLUL, GLYCTK, GM2A, GMNN, GMPPA, GMPPB, GNA11, GNA14, GNAI1, GNAI3, GNAO1, GNAQ, GNAS, GNB1, GNB2, GNB5, GNPAT, GNPTAB, GNPTG, GNS, GOLGA2, GORAB, GOT2, GPC3, GPC4, GPC6, GPSM2, GPT2, GPX4, GPAA1, GREB1L, GRHL2, GRHL3, GRIA1, GRIA2, GRIA3, GRIA4, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM1, GRM6, GRM7, GSPT2, GTF2E2, GTF2H5, GTPBP2, GTPBP3, GUCY2C, GUSB, GZF1, GAA, H1-4, H3-3A, H3-3B, H4C11, H4C3, H4C5, HACD1, HACE1, HADH, HADHA, HAX1, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECTD4, HECW2, HEPACAM, HERC1, HERC2, HESX1, HEXA, HEXB, HGSNAT, HIBCH, HID1, HINT1, HIVEP2, HK1, HLCS, HMGB1, HMGCL, HMGCS2, HMX1, HNF1B, HNF4A, HNMT, HNRNPA2B1, HNRNPD, HNRNPH1, HNRNPH2, HNRNPK, HNRNPR, HNRNPU, HOXA1, HOXA13, HOXB1, HOXC13, HOXD13, HPD, HPDL, HPGD, HPRT1, HPS1, HPSE2, HR, HRAS, HS2ST1, HSD17B10, HSD17B4, HSD3B7, HSF4, HSPD1, HSPG2, HTRA2, HUWE1, HYAL1, HYAL2, HYDIN, HYLS1, HAAO, IARS1, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT74, IFT80, IGF1, IGF1R, IGF2, IGFBP7, IGHMBP2, IGSF1, IHH, IKBKG, IL11RA, IL1RAPL1, IMPDH2, INPP5E, INPP5K, INPPL1, INTS1, INTS11, IPO8, IQSEC1, IQSEC2, IREB2, IRF2BPL, IRF6, IRX5, ITCH, ITGA3, ITGA7, ITGA8, ITPA, ITPR1, IVD, JAG1, JAG2, JAGN1, JAK3, JAM3, JARID2, KANSL1, KARS1, KAT5, KAT6A, KAT6B, KAT8, KATNAL2, KATNB1, KBTBD13, KCNA2, KCNB1, KCNC1, KCNC3, KCND2, KCND3, KCNE1, KCNH1, KCNH5, KCNJ10, KCNJ11, </p> |
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|  | <p> KCNJ6, KCNJ8, KCNK3, KCNK4, KCNK9, KCNMA1, KCNN2, KCNN3, KCNQ1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD1, KCTD3, KCTD7, KDEL2, KDM1A, KDM2B, KDM3B, KDM4B, KDM5A, KDM5B, KDM5C, KDM6A, KDM6B, KIDINS220, KIF11, KIF14, KIF1A, KIF21B, KIF22, KIF2A, KIF4A, KIF5A, KIF5B, KIF5C, KIF7, KIFBP, KIT, KIAA0586, KLF1, KLF7, KLHL20, KLHL40, KLHL7, KMT2A, KMT2B, KMT2C, KMT2D, KMT2E, KMT5B, KNL1, KPTN, KRAS, KRIT1, KRT74, L1CAM, L2HGDH, LAMA1, LAMA2, LAMB1, LAMC3, LAMP2, LARGE1, LARP7, LARS1, LARS2, LBR, LEMD2, LEMD3, LETM1, LFNG, LGI4, LHX2, LHX3, LHX4, LIAS, LIFR, LIG4, LINGO1, LINGO4, LINS1, LIPN, LIPT1, LIPT2, LMBRD1, LMBRD2, LMNA, LMNB1, LMNB2, LMOD3, LMX1B, LNPK, LONP1, LRAT, LRBA, LRIG2, LRIT3, LRP2, LRP4, LRP5, LRPPRC, LRRC56, LSS, LTBP1, LTBP2, LTBP3, LYRM7, LYST, LZTR1, MAB21L1, MAB21L2, MACF1, MADD, MAF, MAFB, MAGEL2, MAMLD1, MAN1B1, MAN2B1, MAN2C1, MANBA, MAOA, MAP1B, MAP2K1, MAP2K2, MAP3K1, MAP3K7, MAPK1, MAPK8IP3, MAPKAPK5, MAPRE2, MASP1, MAST1, MAST4, MAT1A, MATN3, MBD5, MBOAT7, MBTPS2, MC2R, MCCC1, MCCC2, MCEE, MCM3AP, MCOLN1, MCPH1, MDH2, MECOM, MECP2, MECP2, MECP2, MED11, MED12, MED13, MED13L, MED17, MED23, MED25, MED27, MEF2C, MEGF10, MEGF8, MEIS2, MEOX1, MESD, MESP2, METTL23, METTL5, MFF, MFN2, MFRP, MFSD2A, MFSD8, MGAT2, MGP, MICU1, MID1, MINPP1, MIR17HG, MITF, MKKS, MKS1, MLC1, MLYCD, MMAB, MMACHC, MMADHC, MMGT1, MMP13, MMP21, MMUT, MMAA, MN1, MNX1, MOCS1, MOCS2, MOGS, MORC2, MPDU1, MPI, MPLKIP, MPV17, MPZ, MRAS, MRE11, MRPS2, MRPS22, MRPS34, MSL2, MSL3, MSMO1, MSX1, MSX2, MTFMT, MTHFR, MTHFS, MTM1, MTO1, MTOR, MTR, MTRFR, MTRR, MTSS2, MT-TP, MVK, MYBPC1, MYCN, MYF5, MYH10, MYH3, MYH9, MYL11, MYLK, MYO18B, MYO5A, MYO5B, MYO7A, MYOCD, MYPN, MYRF, MYT1L, NACC1, NADSYN1, NAE1, NAGA, NAGLU, NAGS, NALCN, NANS, NAPB, NARS1, NARS2, NAXD, NAXE, NBAS, NBEA, NBN, NCAPG2, NCDN, NCKAP1, NDE1, NDNF, NDP, NDST1, NDUFA1, NDUFA10, NDUFA12, NDUFA2, NDUFA6, NDUFA8, NDUFAF2, NDUFAF8, NDUFB11, NDUFB3, NDUFB8, NDUFS1, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NECTIN4, NEDD4L, NEK1, NEK8, NEMF, NEU1, NEUROD2, NEUROG1, NEXMIF, NF1, NFASC, NFE2L2, NFIA, NFIB, NFIX, NFU1, NGLY1, NHLRC2, NHP2, NHS, NIPBL, NKAP, NKX2-1, NKX2-5, NKX3-2, NKX6-2, NLGN3, NMNAT1, NODAL, NOG, NONO, NOTCH1, NOTCH2, NOVA2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPM1, NPR2, NPR3, NR2F1, NR2F2, NR4A2, NRAS, NRCAM, NRROS, NRXN1, NRXN2, NSD1, NSD2, NSDHL, NSRP1, NSUN2, NT5C2, NT5C3A, NTNG2, NTRK1, NTRK2, NUBPL, NUDT2, NUP133, NUP214, NUP54, NUP62, NUS1, NYX, NAA10, NAA15, OBSL1, OCLN, OCRL, ODAD1, ODAD2, ODAD3, ODAD4, ODAPH, ODC1, OFD1, OGDH, OGDHL, OGT, ONECUT1, OPA3, OPHN1, ORC1, ORC4, ORC6, OSGEP, OTC, OTOGL, OTUD5, OTUD6B, OTUD7A, OTULIN, OTX2, OXCT1, OXR1, P3H1, P4HB, P4HTM, PABPC1, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PAK3, PALB2, PALS1, PAN2, PAPSS2, PARN, PAX1, PAX2, PAX3, PAX6, PAX8, PAX9, PBX1, PC, PCARE, PCBD1, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCGF2, PCNT, PCYT1A, PCYT2, PDCD10, PDE10A, PDE4D, PDE6G, PDE6H, PDGFRB, PDHA1, PDHB, PDHX, PDSS1, PDSS2, PDZD8, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, </p> |
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|  | PGM1, PGM2L1, PGM3, PHACTR1, PHF21A, PHF6, PHF8, PHGDH, PHIP, PHOX2B, PI4KA, PIBF1, PIDD1, PIEZO1, PIEZO2, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY, PIK3CA, PIK3R1, PIK3R2, PIP5K1C, PITRM1, PITX1, PITX2, PITX3, PKD1L1, PKHD1, PLA2G6, PLCB1, PLCB4, PLCE1, PLEC, PLK1, PLK4, PLOD1, PLOD2, PLP1, PLPBP, PLXNA1, PLXND1, PLAA, PMM2, PMPCB, PMS2, PNKP, PNPLA1, PNPLA2, PNPLA6, PNPO, PNPT1, POC1A, POC1B, POGZ, POLA1, POLD1, POLE, POLG, POLR1A, POLR1C, POLR1D, POLR2A, POLR3A, POLR3B, POLRMT, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PORCN, POU1F1, POU3F2, POU3F3, POU4F1, PPA2, PPFIBP1, PPIL1, PPM1D, PPP1CB, PPP1R12A, PPP1R13L, PPP1R15B, PPP1R21, PPP1R3F, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM12, PRDM13, PRDX3, PREPL, PRICKLE2, PRIM1, PRKACA, PRKACB, PRKAR1A, PRKAR1B, PRKD1, PRKG2, PRMT7, PROP1, PRPF8, PRPS1, PRR12, PRRT2, PRSS12, PRSS56, PRUNE1, PSAP, PSAT1, PSMB8, PSMC3, PSMC5, PSMD12, PSPH, PTCH1, PTCHD1, PTDSS1, PTEN, PTF1A, PTH, PTH1R, PTHLH, PTPN11, PTPN14, PTPN23, PTPN4, PTRH2, PTRHD1, PTS, PUF60, PUM1, PURA, PUS1, PUS3, PUS7, PXDN, PYCR1, PYCR2, PYGL, PYROXD1, QARS1, QDPR, QRIC1, RAB11A, RAB11B, RAB14, RAB18, RAB23, RAB39B, RAB3GAP1, RAB3GAP2, RAB5C, RAC1, RAC3, RAD21, RAD51C, RAF1, RAI1, RALA, RALGAPA1, RAP1B, RAPSN, RARB, RARS1, RARS2, RAX, RBBP8, RBL2, RBM10, RBM8A, RBPJ, RBSN, RECQL4, RELN, RERE, REST, RET, RETREG1, RFT1, RFX3, RFX4, RFX6, RFX7, RHOTB2, RIMS2, RIN2, RINT1, RIPK4, RIT1, RLIM, RMND1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF125, RNF13, RNPC3, RNU4ATAC, RNU7-1, ROBO1, ROBO3, ROBO4, ROGDI, ROR2, RORA, RORB, RPE65, RPGRIP1, RPGRIP1L, RPIA, RPL10, RPL11, RPL26, RPS19, RPS23, RPS26, RPS6KA3, RRAS, RRAS2, RRM2B, RSPH1, RSPH3, RSPO2, RSPO4, RSPRY1, RSRC1, RTKL1, RTN4IP1, RTTN, RUNX2, RXYL1, RYR1, SACS, SALL1, SALL4, SAMD9, SAMHD1, SARS1, SARS2, SATB1, SATB2, SBDS, SBF1, SC5D, SCAF4, SCAMP5, SCAPER, SCARF2, SCN11A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCO1, SCO2, SCUBE3, SCYL1, SDCCAG8, SDHA, SDHAF1, SEC23B, SEC24D, SECISBP2, SELENOI, SELENON, SEMA3A, SEMA6B, SEPSECS, SERAC1, SET, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SF3B4, SFXN4, SGPL1, SGSH, SH3PXD2B, SHANK1, SHANK2, SHANK3, SHH, SHMT2, SHOC2, SHOX, SHQ1, SHROOM3, SIAH1, SIK1, SIL1, SIM1, SIN3A, SIN3B, SIX1, SIX3, SIX5, SKI, SKIV2L, SLC10A7, SLC12A2, SLC12A5, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A1, SLC1A2, SLC1A4, SLC22A5, SLC24A4, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A24, SLC25A26, SLC25A38, SLC25A4, SLC25A42, SLC26A2, SLC27A4, SLC2A1, SLC2A10, SLC2A2, SLC30A9, SLC31A1, SLC32A1, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC35D1, SLC37A4, SLC38A3, SLC39A13, SLC39A14, SLC39A8, SLC45A1, SLC46A1, SLC4A1, SLC4A11, SLC4A4, SLC52A2, SLC52A3, SLC5A5, SLC5A6, SLC5A7, SLC6A1, SLC6A17, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC9A6, SLF2, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCA5, SMARCA11, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMC5, SMCHD1, SMG8, SMG9, SMO, SMOC1, SMOC2, SMPD1, SMPD4, SMS, SNAP25, SNAP29, SNIP1, SNORD118, SNRPB, SNRPE, SNX14, SNX27, SON, SOS1, SOS2, SOX10, SOX11, SOX17, SOX2, SOX3, SOX4, SOX5, SOX6, SOX9, SPAG1, SPARC, SPART, SPAST, SPATA5, SPATA5L1, |
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|  | <p> SPECC1L, SPEG, SPEN, SPG11, SPOP, SPR, SPRED1, SPRED2, SPTAN1, SPTBN1, SPTBN2, SPTBN4, SPTLC2, SRCAP, SRD5A3, SRP54, SRRM2, SRSF1, SRY, SSR4, ST14, ST3GAL3, ST3GAL5, STAC3, STAG1, STAG2, STAMBP, STAR, STAT5B, STEEP1, STIL, STN1, STRA6, STRADA, STS, STT3A, STX1B, STXBP1, SUCLG1, SUFU, SUMF1, SUOX, SUPT16H, SURF1, SUZ12, SVBP, SYN1, SYNCRIP, SYNE1, SYNGAP1, SYNJ1, SYP, SYT1, SYT2, SZT2, TAB2, TAC3, TACO1, TACR3, TAF1, TAF13, TAF2, TAF4, TAF6, TAF8, TANC2, TANGO2, TAOK1, TAPT1, TARS1, TASP1, TAT, TBC1D20, TBC1D23, TBC1D24, TBC1D2B, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TBX15, TBX18, TBX20, TBX22, TBX3, TBX4, TBX5, TBXAS1, TCEAL1, TCF12, TCF20, TCF4, TCF7L2, TCN2, TCOF1, TCTN1, TCTN2, TCTN3, TDP2, TECPR2, TEFM, TEK, TELO2, TENM3, TERC, TERT, TET3, TFAP2A, TFAP2B, TFE3, TGDS, TGFB1, TGFB2, TGFB3, TGFB1, TGFB2, TGIF1, TH, THAP1, THOC2, THOC6, THRA, THUMPD1, TIAM1, TIMM50, TIMM8A, TINF2, TK2, TKT, TLK2, TMC01, TMEM106B, TMEM126B, TMEM147, TMEM163, TMEM165, TMEM199, TMEM216, TMEM218, TMEM222, TMEM237, TMEM240, TMEM251, TMEM260, TMEM63A, TMEM63B, TMEM63C, TMEM67, TMEM70, TMEM94, TMPRSS6, TMTC3, TMX2, TNFRSF13B, TNNT3, TNPO2, TNRC6B, TOE1, TOGARAM1, TOP3A, TOR1A, TP53RK, TP63, TP73, TPM2, TPM3, TPP1, TPP2, TRA2B, TRAF7, TRAI, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC4, TRAPPC6B, TRAPPC9, TREX1, TRIM32, TRIM37, TRIM8, TRIO, TRIP11, TRIP12, TRIP13, TRIP4, TRIT1, TRMT1, TRMT10A, TRMT10C, TRNT1, TRPM1, TRPM3, TRPS1, TRPV3, TRPV4, TRPV6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TSPOAP1, TTC19, TTC37, TTC5, TTC7A, TTC8, TTI1, TTI2, TTN, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, TUFM, TUSC3, TWIST1, TWIST2, TXNL4A, TYR, TYRP1, U2AF2, UBA5, UBAP2L, UBE2A, UBE2T, UBE3A, UBE3B, UBE4A, UBR1, UBR7, UBTF, UFC1, UFM1, UFSP2, UGDH, UGP2, UGT1A1, UMPS, UNC45A, UNC45B, UNC80, UPF1, UPF3B, UQCRB, UQCRRF1, UQCRRQ, UROC1, UROS, USB1, USP18, USP27X, USP7, USP9X, UVSSA, VAMP2, VARS1, VARS2, VCP, VDR, VIPAS39, VLDLR, VPS11, VPS13B, VPS33B, VPS41, VPS4A, VPS53, VRK1, VSX2, WAC, WARS1, WARS2, WASF1, WASHC5, WDFY3, WDPCP, WDR11, WDR19, WDR26, WDR35, WDR37, WDR4, WDR45, WDR45B, WDR5, WDR62, WDR73, WDR81, WFS1, WIPI2, WNK3, WNT1, WNT10B, WNT3, WNT4, WNT5A, WNT7A, WRAP53, WT1, WWOX, WWTR1, XPA, XPC, XRCC4, XYLT1, XYLT2, YAP1, YARS2, YIF1B, YIPF5, YRDC, YWHAG, YY1, ZBTB18, ZBTB20, ZBTB24, ZBTB47, ZBTB7A, ZC4H2, ZDHHC9, ZEB2, ZFHX4, ZFP57, ZFYVE19, ZFYVE26, ZIC1, ZIC2, ZIC3, ZMIZ1, ZMPSTE24, ZMYM2, ZMYM3, ZMYND10, ZMYND11, ZMYND8, ZNF142, ZNF148, ZNF292, ZNF335, ZNF462, ZNF526, ZNF699, ZNF711, ZNF750, ZSWIM6, AAAS, AARS1, AASS. </p> |
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| Primary immunodeficiency    | <p>ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AGR2, AICDA, AIRE, AK2, ALPI, ALPK1, AP3B1, ARPC1B, ARPC5, ATM, ATP6AP1, B2M, BACH2, BCL10, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C2orf69, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CBLB, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CDC42, CDCA7, CEBPE, CFD, CFH, CFI, CFP, CHD7, CIB1, CIITA, CLPB, COL7A1, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTLA4, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DBR1, DCLRE1C, DEF6, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DUT, EFL1, ELANE, ELF4, EPCAM, EPG5, ERCC6L2, EXTL3, F12, FADD, FAS, FASLG, FAT4, FCHO1, FERMT1, FERMT3, FNIP1, FOXP1, FOXP3, G6PC3, G6PD, GATA1, GATA2, GFI1, GINS1, GUCY2C, HAVCR2, HAX1, HELLS, HMOX1, HPS1, HPS4, HPS6, HTRA2, HYOU1, ICOS, IFIH1, IFNAR2, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, IRAK4, IRF2BP2, IRF4, IRF7, IRF8, ISG15, ITCH, ITGB2, ITK, JAGN1, JAK1, JAK3, KDM6A, KMT2A, KMT2D, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LYN, LYST, MAGT1, MALT1, MAP3K14, MCM4, MCTS1, MECOM, MEFV, MOGS, MPEG1, MSN, MTHFD1, MVK, MYD88, MYO5B, MYSM1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NPC1, NSMCE3, OAS1, ORAI1, OTULIN, PARN, PAX1, PEPD, PGM3, PI4KA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PLG, PNP, POLA1, POLD1, POLE, POMP, PRF1, PRIM1, PRKCD, PRKDC, PSMB10, PSMB4, PSMB8, PSMB9, PSTPIP1, PTPN2, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RECQL4, REL, RELA, RFX5, RFXANK, RFXAP, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNU7-1, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SEC61A1, SERPING1, SGPL1, SH2D1A, SH3BP2, SKIV2L, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SLCO2A1, SMARCA1, SMARCD2, SNORA31, SOCS1, SP110, SPI1, SPINK5, SPPL2A, SRP54, STAT1, STAT2, STAT3, STAT5B, STAT6, STIM1, STING1, STK4, STX11, STXBP2, STXBP3, SYK, TAP1, TAP2, TBK1, TBX1, TCF3, TCN2, TFRC, TGFB1, TGFB1R1, TGFB1R2, TICAM1, TLR3, TLR7, TLR8, TMC6, TMC8, TNFAIP3, TNFRSF1A, TNFRSF9, TOP2B, TPP2, TRAC, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC37, TTC7A, TYK2, UBA1, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, WWTR1, XIAP, ZAP70, ZBTB24, ZNF341, ZNF1.</p> |
| Cholestasis (Miscellaneous) | <p>ABCB11, ABCB4, ABCC2, ADK, AKR1D1, ALDOB, AMACR, ATP7B, ATP8B1, BAAT, BCS1L, CFTR, CLDN1, COG7, CYP27A1, CYP7B1, DCDC2, DGUOK, FAH, GALE, GALM, GALT, GBA, HADHA, HNF1B, HSD3B7, JAG1, KIF12, LIPA, MPI, MPV17, MVK, MYO5B, NBAS, NOTCH2, NPC1, NPC2, NR1H4, PEX1, PEX12, PEX26, PEX6, PKHD1, POLG, RINT1, SERPINA1, SLC25A13, SMPD1, TALDO1, TJP2, TRMU, UGT1A1, UNC45A, USP53, VIPAS39, VPS33B, YARS1, ZFYVE19.</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              |

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|---------------------------------|------------------------------|
| Pancreatitis<br>(Miscellaneous) | CELA3B, CFTR, PRSS1, SPINK1. |
|---------------------------------|------------------------------|

**Table S3: Genes contained in the applied reanalysis *in silico* gene panels**

A final merged *in silico* gene panel was created per patient group, except for hematological disease and miscellaneous, to be used for reanalysis. The size of the applied WGS reanalysis *in silico* gene panels ranged from 4 genes (Pancreatitis) to 2,259 genes (Neurodevelopmental disorders).