



Extended Gene List  
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## Gene & condition list

Please note this gene list is a subset of 629 genes from the Lumi Comprehensive Carrier Screening gene list.

Please see the [Lumi Comprehensive Test gene and condition list](#) for more information on some of the genes below.

| Genes   |          |        |
|---------|----------|--------|
| AAAS    | ADGRG1   | ALDOB  |
| AARS2   | ADGRV1   | ALG1   |
| ABCA12  | ADSL     | ALG3   |
| ABCA3   | AGA      | ALG6   |
| ABCB11  | AGK      | ALMS1  |
| ABCB4   | AGL      | ALPL   |
| ABCC8   | AGPS     | AMPD2  |
| ABCD1   | AGXT     | AMT    |
| ACAD9   | AHI1     | AP1S2  |
| ACADM   | AIFM1    | AQP2   |
| ACADVL  | AIPL1    | ARG1   |
| ACAT1   | AK2      | ARL13B |
| ACOX1   | ALDH18A1 | ARL6   |
| ADA     | ALDH3A2  | ARSA   |
| ADAMTS2 | ALDH5A1  | ARSB   |
| ADAR    | ALDH7A1  | ARX    |
| ASL     | BBS9     | CD40   |
| ASNS    | BCKDHA   | CD40LG |
| ASPA    | BCKDHB   | CDH23  |
| ASPM    | BCS1L    | CENPJ  |

|          |         |        |
|----------|---------|--------|
| ASS1     | BLM     | CEP152 |
| ATM      | BRAT1   | CEP290 |
| ATP6V1B1 | BRWD3   | CEP41  |
| ATP7A    | BSND    | CFTR   |
| ATP7B    | BTK     | CHAT   |
| ATP8B1   | CANT1   | CHRNE  |
| ATR      | CAPN3   | CHRNA  |
| ATRX     | CASK    | CIITA  |
| AUH      | CASQ2   | CKAP2L |
| B3GLCT   | CC2D1A  | CLCN5  |
| BBS1     | CC2D2A  | CLCN7  |
| BBS10    | CCBE1   | CLN3   |
| BBS12    | CCDC103 | CLN5   |
| BBS2     | CCDC39  | CLN6   |
| BBS4     | CCDC88C | CLN8   |
| BBS5     | CCN6    | CLP1   |
| BBS7     | CD3D    | CLPB   |
| CLRN1    | CTNS    | DGAT1  |
| CNGB3    | CTSA    | DGUOK  |
| COL11A2  | CTSC    | DHCR24 |
| COL17A1  | CTSD    | DHCR7  |
| COL18A1  | CTSK    | DHDDS  |
| COL27A1  | CUL4B   | DIS3L2 |
| COL4A3   | CYBA    | DKC1   |
| COL4A4   | CYBB    | DLD    |
| COL4A5   | CYP11A1 | DLG3   |

|         |         |         |
|---------|---------|---------|
| COL6A1  | CYP11B2 | DLL3    |
| COL7A1  | CYP17A1 | DMD     |
| COLEC11 | CYP1B1  | DNAH11  |
| COLQ    | CYP27A1 | DNAH5   |
| COX15   | CYP7B1  | DNAI1   |
| CPLANE1 | D2HGDH  | DNAI2   |
| CPS1    | DBT     | DNMT3B  |
| CPT1A   | DCAF17  | DOCK6   |
| CPT2    | DCLRE1C | DOK7    |
| CRB1    | DCX     | DYM     |
| CRTAP   | DDC     | DYNC2H1 |
| CSPP1   | DDX11   | DYNC2I2 |
| DYSF    | ETFDH   | FH      |
| ECHS1   | ETHE1   | FHL1    |
| EDA     | EVC     | FKBP10  |
| EIF2AK3 | EVC2    | FKRP    |
| EIF2B1  | EXOSC3  | FKTN    |
| EIF2B2  | EXOSC8  | FLNA    |
| EIF2B3  | F2      | FMR1    |
| EIF2B4  | FAH     | FOXN1   |
| EIF2B5  | FAM126A | FOXRED1 |
| ELP1    | FANCA   | FRAS1   |
| EMD     | FANCB   | FREM2   |
| ENPP1   | FANCC   | FTSJ1   |
| EPG5    | FANCD2  | FUCA1   |
| ERCC2   | FANCE   | G6PC    |

|        |        |          |
|--------|--------|----------|
| ERCC4  | FANCF  | G6PC3    |
| ERCC5  | FANCG  | GAA      |
| ERCC6  | FANCI  | GALC     |
| ERCC8  | FANCL  | GALNS    |
| ESCO2  | FAT4   | GALT     |
| ETFA   | FBP1   | GAMT     |
| ETFB   | FBXO7  | GATM     |
| GBE1   | GPR143 | HPS1     |
| GCDH   | GPSM2  | HPS3     |
| GCH1   | GSS    | HPS4     |
| GDAP1  | GUCY2D | HPS5     |
| GDF1   | GUSB   | HPS6     |
| GDF5   | HADH   | HSD17B10 |
| GFM1   | HADHA  | HSD17B4  |
| GHR    | HADHB  | HSD3B2   |
| GJB1   | HAMP   | HUWE1    |
| GLA    | HAX1   | HYLS1    |
| GLB1   | HBB    | IDS      |
| GLDC   | HCFC1  | IDUA     |
| GLE1   | HEXA   | IGHMBP2  |
| GNB5   | HEXB   | IKBKB    |
| GNE    | HGSNAT | IL1RAPL1 |
| GNPAT  | HIBCH  | IL2RG    |
| GNPTAB | HLCS   | IL7R     |
| GNPTG  | HMGCL  | INPP5E   |
| GNS    | HMGCS2 | INVS     |

|        |         |         |
|--------|---------|---------|
| GORAB  | HPD     | IQSEC2  |
| GPC3   | HPRT1   | ITGA6   |
| ITGB4  | LARS1   | MECP2   |
| ITPR1  | LCA5    | MED12   |
| IVD    | LDLR    | MED17   |
| JAK3   | LDLRAP1 | MESP2   |
| KATNB1 | LHX3    | METTL23 |
| KCNJ1  | LIFR    | MFN2    |
| KCNJ11 | LIG4    | MFSD8   |
| KCNQ1  | LIPA    | MID1    |
| KDM5C  | LMBRD1  | MKKS    |
| KIF1A  | LMNA    | MKS1    |
| KIF7   | LPL     | MLC1    |
| KRT14  | LRAT    | MLYCD   |
| L1CAM  | LRP2    | MMAA    |
| L2HGDH | LRPPRC  | MMAB    |
| LAMA2  | LYST    | MMACHC  |
| LAMA3  | LZTFL1  | MMADHC  |
| LAMB1  | MAN2B1  | MOCS1   |
| LAMB2  | MANBA   | MOCS2   |
| LAMB3  | MASP1   | MPI     |
| LAMC2  | MCOLN1  | MPL     |
| LARGE1 | MCPH1   | MPV17   |
| MRE11  | NDRG1   | OPA1    |
| MTFMT  | NDUFAF2 | OPA3    |
| MTHFR  | NDUFAF5 | OPHN1   |

|       |        |          |
|-------|--------|----------|
| MTM1  | NDUFS4 | OSGEP    |
| MTMR2 | NDUFS6 | OSTM1    |
| MTR   | NDUFS7 | OTC      |
| MTRR  | NDUFV1 | P3H1     |
| MTTP  | NEB    | PAH      |
| MUSK  | NEU1   | PAK3     |
| MVK   | NGLY1  | PANK2    |
| MYO5B | NNT    | PC       |
| MYO7A | NPC1   | PCCA     |
| NAGA  | NPC2   | PCCB     |
| NAGLU | NPHP1  | PCDH15   |
| NAGS  | NPHP3  | PCDH19   |
| NALCN | NPHS1  | PCNT     |
| NARS2 | NPHS2  | PDHA1    |
| NBN   | NR0B1  | PDHB     |
| NCF2  | NTRK1  | PEPD     |
| NDE1  | OCRL   | PET100   |
| NDP   | OFD1   | PEX1     |
| PEX10 | PKHD1  | PRPS1    |
| PEX12 | PLA2G6 | PSAP     |
| PEX13 | PLOD1  | PTS      |
| PEX16 | PLP1   | PUS1     |
| PEX2  | PLPBP  | QDPR     |
| PEX26 | PMM2   | RAB18    |
| PEX5  | PNKP   | RAB23    |
| PEX6  | PNPO   | RAB3GAP1 |

|          |          |          |
|----------|----------|----------|
| PEX7     | POLG     | RAB3GAP2 |
| PFKM     | POLR1C   | RAG1     |
| PGAP2    | POLR3B   | RAG2     |
| PGK1     | POMGNT1  | RAPSN    |
| PGM1     | POMT1    | RARS2    |
| PGM3     | POMT2    | RAX      |
| PHF8     | POR      | RBBP8    |
| PHGDH    | POU1F1   | RDH12    |
| PHYH     | PPT1     | RMND1    |
| PIBF1    | PQBP1    | RMRP     |
| PIGG     | PRDM5    | RNASEH2A |
| PIGN     | PRF1     | RNASEH2B |
| PIGT     | PROP1    | RNASEH2C |
| RP2      | SKIV2L   | SLC52A3  |
| RPE65    | SLC12A1  | SLC6A5   |
| RPGRIP1L | SLC12A6  | SLC6A8   |
| RPS6KA3  | SLC16A2  | SLC7A7   |
| RTEL1    | SLC17A5  | SMARCAL1 |
| RYR1     | SLC19A2  | SMN1     |
| SACS     | SLC19A3  | SMPD1    |
| SAMHD1   | SLC1A4   | SNAP29   |
| SC5D     | SLC22A5  | SPATA5   |
| SCO2     | SLC25A1  | SPG11    |
| SDCCAG8  | SLC25A13 | SPINK5   |
| SEC23B   | SLC25A15 | SPR      |
| SEPSECS  | SLC26A2  | ST3GAL5  |



|          |         |        |
|----------|---------|--------|
| SERAC1   | SLC26A3 | STAR   |
| SERPINH1 | SLC35A3 | STX11  |
| SGCA     | SLC37A4 | STXBP2 |
| SGCB     | SLC38A8 | SUMF1  |
| SGCD     | SLC39A4 | SUOX   |
| SGCG     | SLC45A2 | SURF1  |
| SGSH     | SLC46A1 | SYN1   |
| SH3TC2   | SLC52A2 | TANGO2 |
| TAT      | TMEM67  | TYR    |
| TAZ      | TMTC3   | TYRP1  |
| TBC1D23  | TOE1    | UBA5   |
| TBC1D24  | TPP1    | UBE2T  |
| TBCD     | TRDN    | UBR1   |
| TBCE     | TREX1   | UGT1A1 |
| TCIRG1   | TRIM32  | UNC13D |
| TCN2     | TRIM37  | UPF3B  |
| TCTN2    | TRMU    | USH1C  |
| TCTN3    | TRPM6   | USH1G  |
| TECPR2   | TSEN2   | USH2A  |
| TELO2    | TSEN54  | USP9X  |
| TF       | TSMF    | VLDLR  |
| TGM1     | TSHB    | VPS11  |
| TH       | TTC37   | VPS13B |
| THOC2    | TTC7A   | VPS45  |
| TK2      | TTC8    | VPS53  |
| TMEM138  | TTPA    | VRK1   |

|         |        |         |
|---------|--------|---------|
| TMEM216 | TULP1  | VSX2    |
| TMEM231 | TWNK   | WAS     |
| TMEM237 | TYMP   | WDR62   |
| WDR81   | XPA    | ZFYVE26 |
| WHRN    | XPC    | ZNF711  |
| WRN     | YARS2  | HFE2    |
| WWOX    | ZBTB24 | MUT     |
| XIAP    | ZDHHC9 |         |