

Whole Exome Sequencing

Gene package Primary Immunodeficiency Disorders, version 9, 26-2-2021



Technical information

DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: <http://bio-bwa.sourceforge.net/>). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: <http://www.broadinstitute.org/gatk/>). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Copy variant detection is performed using the BAM multiscale reference method using depth of coverage analysis and dynamical bins in NexusClinical. The detected copy number variants are filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). It is not excluded that pathogenic variants are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).



Dept. Clinical Genetics

| HGNC approved gene symbol | OMIM gene ID (active link to omim.org) | % covered $\geq 10x$ | % covered $\geq 20x$ | % covered $\geq 30x$ | % covered $\geq 50x$ |
|---------------------------|---|----------------------|----------------------|----------------------|----------------------|
| ACD | 609377 | 100 | 100 | 100 | 94.73 |
| ACP5 | 171640 | 100 | 100 | 100 | 100 |
| ACTB | 102630 | 100 | 100 | 100 | 98.33 |
| ADA | 608958 | 100 | 100 | 99.88 | 92.24 |
| ADA2 | 607575 | 100 | 100 | 100 | 99.95 |
| ADAM17 | 603639 | 100 | 98.56 | 95.42 | 88.36 |
| ADAMTS3 | 605011 | 100 | 100 | 99.99 | 99.16 |
| ADAR | 146920 | 98.45 | 98.45 | 98.45 | 97.45 |
| AGA | 613228 | 100 | 100 | 98.65 | 89.74 |
| AICDA | 605257 | 100 | 100 | 99.57 | 81.63 |
| AIRE | 607358 | 100 | 99.35 | 95.72 | 87.42 |
| AK2 | 103020 | 100 | 99.22 | 95.40 | 86.92 |
| ALG13 | 300776 | 100 | 100 | 99.14 | 87.52 |
| ALPI | 171740 | 100 | 100 | 100 | 97.39 |
| ANGPT1 | 601667 | 100 | 100 | 100 | 99.19 |
| AP1S3 | 615781 | 100 | 100 | 96.17 | 95.91 |
| AP3B1 | 603401 | 97.72 | 95.91 | 94.30 | 84.26 |
| AP3D1 | 607246 | 97.71 | 97.16 | 95.44 | 86.83 |
| APOL1 | 603743 | 100 | 100 | 100 | 97.94 |
| ARHGEF1 | 601855 | 100 | 99.12 | 95.85 | 80.17 |
| ARPC1B | 604223 | 100 | 100 | 100 | 95.56 |
| ATM | 607585 | 100 | 94.70 | 74.49 | 49.62 |
| ATP6AP1 | 300197 | 100 | 99.75 | 95.65 | 86.21 |
| B2M | 109700 | 100 | 100 | 100 | 100 |

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|---------------------------|--|----------------|----------------|----------------|----------------|
| BACH2 | 605394 | 100 | 100 | 100 | 98.21 |
| BCL10 | 603517 | 100 | 99.01 | 89.86 | 85.70 |
| BCL11B | 606558 | 100 | 98.43 | 96.33 | 84.36 |
| BLK | 191305 | 100 | 99.26 | 96.01 | 85.64 |
| BLM | 604610 | 100 | 98.83 | 97.55 | 91.70 |
| BLNK | 604515 | 99.47 | 96.05 | 92.97 | 85.48 |
| BLOC1S6 | 604310 | 100 | 94.61 | 78.45 | 78.29 |
| BTK | 300300 | 100 | 100 | 99.69 | 93.31 |
| C1QA | 120550 | 100 | 100 | 100 | 100 |
| C1QB | 120570 | 100 | 100 | 100 | 97.00 |
| C1QC | 120575 | 100 | 100 | 100 | 93.87 |
| C1R | 613785 | 100 | 100 | 99.24 | 94.22 |
| C1S | 120580 | 100 | 100 | 99.06 | 94.05 |
| C2 | 613927 | 100 | 100 | 100 | 95.13 |
| C3 | 120700 | 100 | 100 | 99.09 | 94.06 |
| C5 | 120900 | 99.93 | 99.16 | 96.69 | 90.42 |
| C6 | 217050 | 100 | 100 | 100 | 99.22 |
| C7 | 217070 | 100 | 100 | 100 | 99.08 |
| C8A | 120950 | 100 | 100 | 100 | 97.69 |
| C8B | 120960 | 100 | 100 | 100 | 98.34 |
| C8G | 120930 | 100 | 100 | 94.64 | 74.66 |
| C9 | 120940 | 100 | 100 | 100 | 97.89 |
| CA2 | 611492 | 100 | 100 | 100 | 97.88 |
| CARD11 | 607210 | 100 | 99.12 | 97.01 | 92.44 |
| CARD14 | 607211 | 100 | 98.89 | 96.67 | 86.80 |
| CARD9 | 607212 | 100 | 98.97 | 97.00 | 86.44 |
| CARMIL2 | 610859 | 97.81 | 94.63 | 92.52 | 82.62 |
| CASP10 | 601762 | 100 | 100 | 100 | 100 |
| CASP8 | 601763 | 100 | 98.88 | 95.53 | 91.74 |
| CAVIN1 | 603198 | 100 | 100 | 100 | 95.50 |
| CCBE1 | 612753 | 100 | 100 | 99.58 | 94.92 |
| CD19 | 107265 | 100 | 99.56 | 97.49 | 90.21 |
| CD247 | 186780 | 100 | 100 | 100 | 96.63 |
| CD27 | 186711 | 100 | 100 | 100 | 95.78 |
| CD3D | 186790 | 100 | 100 | 100 | 100 |
| CD3E | 186830 | 100 | 100 | 100 | 94.56 |
| CD3G | 186740 | 100 | 100 | 100 | 100 |
| CD40 | 109535 | 100 | 100 | 100 | 98.05 |
| CD40LG | 300386 | 100 | 100 | 100 | 96.60 |
| CD46 | 120920 | 100 | 100 | 100 | 95.54 |
| CD55 | 125240 | 100 | 96.43 | 91.53 | 78.51 |
| CD59 | 107271 | 100 | 100 | 100 | 100 |
| CD70 | 602840 | 100 | 100 | 100 | 88.78 |
| CD79A | 112205 | 100 | 91.32 | 83.16 | 62.79 |

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|---------------------------|--|----------------|----------------|----------------|----------------|
| CD79B | 147245 | 100 | 100 | 100 | 99.51 |
| CD81 | 186845 | 100 | 100 | 96.89 | 81.11 |
| CD8A | 186910 | 100 | 100 | 99.27 | 86.61 |
| CDCA7 | 609937 | 100 | 98.06 | 93.10 | 83.32 |
| CDKN2B | 600431 | 100 | 100 | 100 | 91.96 |
| CEBPE | 600749 | 100 | 100 | 100 | 100 |
| CFB | 138470 | 100 | 100 | 100 | 98.76 |
| CFD | 134350 | 100 | 99.94 | 88.81 | 67.56 |
| CFH | 134370 | 99.46 | 97.06 | 94.46 | 90.27 |
| CFHR1 | 134371 | 97.66 | 92.97 | 89.32 | 82.79 |
| CFHR3 | 605336 | 100 | 99.89 | 90.35 | 71.94 |
| CFHR5 | 608593 | 100 | 98.53 | 95.25 | 88.54 |
| CFI | 217030 | 98.47 | 95.86 | 92.94 | 91.23 |
| CFP | 300383 | 100 | 99.81 | 97.95 | 88.19 |
| CFTR | 602421 | 100 | 100 | 99.58 | 95.66 |
| CHD7 | 608892 | 100 | 99.87 | 98.61 | 96.80 |
| CIB1 | 602293 | 100 | 100 | 98.20 | 87.82 |
| CIITA | 600005 | 100 | 99.24 | 96.46 | 84.40 |
| CLCN7 | 602727 | 100 | 100 | 99.30 | 89.11 |
| CLEC4D | 609964 | 100 | 100 | 100 | 93.27 |
| CLEC7A | 606264 | 100 | 100 | 100 | 96.75 |
| CLPB | 616254 | 100 | 100 | 100 | 98.07 |
| COG6 | 606977 | 100 | 100 | 99.53 | 92.92 |
| COPA | 601924 | 100 | 100 | 99.61 | 96.71 |
| CORO1A | 605000 | 92.29 | 91.91 | 90.49 | 85.72 |
| CR2 | 120650 | 100 | 100 | 99.58 | 95.80 |
| CREBBP | 600140 | 99.64 | 98.59 | 96.38 | 88.55 |
| CSF2RA | 306250 | 100 | 100 | 100 | 98.34 |
| CSF2RB | 425000 | 100 | 98.75 | 96.09 | 86.72 |
| CSF3R | 138981 | 100 | 98.39 | 94.83 | 85.17 |
| CTC1 | 138971 | 100 | 100 | 99.31 | 90.43 |
| CTLA4 | 613129 | 100 | 100 | 100 | 99.33 |
| CTPS1 | 123890 | 100 | 100 | 99.01 | 93.87 |
| CTSC | 123860 | 100 | 100 | 100 | 100 |
| CXCR4 | 602365 | 100 | 100 | 99.40 | 93.23 |
| CYBA | 162643 | 94.68 | 83.76 | 65.53 | 50.71 |
| CYBB | 608508 | 100 | 99.95 | 97.03 | 83.25 |
| CYBC1 | 300481 | 100 | 96.84 | 89.21 | 83.55 |
| DBR1 | 607024 | 100 | 100 | 100 | 96.15 |
| DCLRE1B | 609683 | 100 | 100 | 100 | 95.27 |
| DCLRE1C | 605988 | 100 | 100 | 99.04 | 90.85 |
| DDX58 | 609631 | 100 | 100 | 98.96 | 95.47 |
| DEF6 | 610094 | 100 | 96.55 | 92.66 | 87.20 |
| DGAT1 | 604900 | 95.59 | 90.52 | 87.80 | 86.72 |

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|---------------------------|--|----------------|----------------|----------------|----------------|
| DHFR | 126060 | 100 | 100 | 98.31 | 83.92 |
| DKC1 | 300126 | 100 | 98.05 | 94.16 | 83.85 |
| DNAJC21 | 617048 | 100 | 99.04 | 95.85 | 81.46 |
| DNASE1 | 125505 | 100 | 100 | 100 | 95.53 |
| DNASE1L3 | 602244 | 100 | 100 | 100 | 96.68 |
| DNASE2 | 126350 | 100 | 100 | 96.58 | 86 |
| DNMT3B | 602900 | 100 | 99.83 | 98.20 | 90.78 |
| DOCK2 | 603122 | 100 | 99.54 | 98.91 | 95.43 |
| DOCK8 | 611432 | 100 | 99.53 | 98.26 | 93.32 |
| EFL1 | 617538 | 100 | 100 | 100 | 98.37 |
| ELANE | 130130 | 100 | 100 | 100 | 95.67 |
| ELF4 | 300775 | 100 | 99.86 | 98.53 | 92.18 |
| EPG5 | 615068 | 100 | 99.62 | 98.57 | 93.31 |
| ERCC2 | 126340 | 100 | 100 | 99.69 | 90.71 |
| ERCC3 | 133510 | 100 | 100 | 99.96 | 97.34 |
| ERCC6L2 | 615667 | 100 | 100 | 99.34 | 93.43 |
| EXTL3 | 605744 | 100 | 100 | 100 | 100 |
| F12 | 610619 | 100 | 100 | 100 | 97.11 |
| FAAP24 | 610884 | 100 | 100 | 100 | 94.14 |
| FADD | 602457 | 100 | 100 | 100 | 99.92 |
| FAS | 134637 | 100 | 100 | 99.63 | 87.44 |
| FASLG | 134638 | 100 | 100 | 96.48 | 87.70 |
| FAT4 | 612411 | 100 | 99.99 | 99.92 | 99.24 |
| FCGR1A | 146760 | 100 | 96.70 | 96.70 | 96.22 |
| FCGR2A | 146790 | 100 | 100 | 100 | 98.99 |
| FCGR2B | 604590 | 76.70 | 71.83 | 71.83 | 66.10 |
| FCGR3A | 146740 | 100 | 100 | 100 | 98.39 |
| FCGR3B | 610665 | 100 | 100 | 100 | 100 |
| FCHO1 | 613437 | 100 | 98.00 | 92.67 | 77.84 |
| FCN3 | 604973 | 100 | 100 | 99.57 | 83.68 |
| FERMT3 | 607901 | 100 | 98.11 | 93.07 | 84.17 |
| FNIP1 | 610594 | 100 | 99.82 | 99.08 | 96.10 |
| FOXP3 | 300292 | 100 | 100 | 99.17 | 89.13 |
| FPR1 | 136537 | 100 | 100 | 100 | 100 |
| G6PC | 613742 | 100 | 100 | 100 | 100 |
| G6PC3 | 611045 | 100 | 93.28 | 85.47 | 65.26 |
| G6PD | 305900 | 100 | 100 | 100 | 94.62 |
| GATA2 | 137295 | 100 | 100 | 99.97 | 80.65 |
| GFI1 | 600871 | 100 | 99.57 | 98.12 | 91.23 |
| GIN51 | 610608 | 100 | 100 | 100 | 100 |
| GJC2 | 608803 | 98.92 | 91.29 | 81.49 | 61.37 |
| GRHL2 | 608576 | 100 | 100 | 99.09 | 92.21 |
| GTF2H5 | 608780 | 100 | 100 | 100 | 100 |

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|---------------------------|--|----------------|----------------|----------------|----------------|
| HAVCR2 | 606652 | 100 | 100 | 100 | 93.72 |
| HAX1 | 605998 | 100 | 100 | 100 | 98.41 |
| HELLS | 603946 | 100 | 97.53 | 93.93 | 86.21 |
| HMOX1 | 141250 | 100 | 100 | 98.34 | 89.52 |
| HYOU1 | 601746 | 100 | 100 | 98.46 | 94.38 |
| ICOS | 604558 | 100 | 100 | 100 | 100 |
| ICOSLG | 605717 | 100 | 100 | 100 | 100 |
| IFIH1 | 606951 | 100 | 100 | 100 | 97.67 |
| IFNAR1 | 107450 | 100 | 100 | 98.36 | 88.95 |
| IFNAR2 | 602376 | 100 | 100 | 100 | 97.26 |
| IFNG | 147570 | 99.83 | 99.83 | 99.83 | 95.16 |
| IFNGR1 | 107470 | 100 | 99.32 | 93.65 | 90.48 |
| IFNGR2 | 147569 | 97.27 | 92.43 | 92.43 | 87.62 |
| IGLL1 | 146770 | 100 | 100 | 99.72 | 87.18 |
| IKBKB | 603258 | 100 | 99.81 | 98.47 | 90.57 |
| IKBKG | 300248 | 33.32 | 25.95 | 25.95 | 22.46 |
| IKZF1 | 603023 | 100 | 100 | 99.08 | 94.82 |
| IL10 | 124092 | 100 | 100 | 100 | 99.13 |
| IL10RA | 146933 | 100 | 100 | 98.56 | 94.08 |
| IL10RB | 123889 | 100 | 94.66 | 93.72 | 89.42 |
| IL12B | 161561 | 100 | 100 | 98.78 | 87.82 |
| IL12RB1 | 601604 | 100 | 99.74 | 96.61 | 81.71 |
| IL17F | 606496 | 100 | 100 | 100 | 98.18 |
| IL17RA | 605461 | 100 | 99.70 | 94.47 | 88.33 |
| IL17RC | 610925 | 100 | 98.09 | 93.79 | 78.97 |
| IL18BP | 604113 | 100 | 100 | 100 | 96.85 |
| IL1RN | 147679 | 100 | 100 | 100 | 98.94 |
| IL2 | 147680 | 100 | 100 | 97.96 | 91.00 |
| IL21 | 605384 | 100 | 99.15 | 92.92 | 83.28 |
| IL21R | 605383 | 100 | 100 | 100 | 95.94 |
| IL2RA | 147730 | 100 | 100 | 100 | 98.51 |
| IL2RB | 146710 | 100 | 100 | 98.31 | 90.97 |
| IL2RG | 308380 | 100 | 100 | 99.61 | 90.29 |
| IL36RN | 605507 | 100 | 100 | 97.61 | 83.67 |
| IL6R | 147880 | 100 | 98.50 | 92.52 | 82.01 |
| IL6ST | 600694 | 100 | 99.67 | 96.71 | 91.70 |
| IL7R | 146661 | 100 | 100 | 100 | 100 |
| INO80 | 610169 | 100 | 99.50 | 98.58 | 93.75 |
| INSR | 147670 | 100 | 99.13 | 98.14 | 96.23 |
| IRAK1 | 300283 | 99.83 | 97.76 | 91.72 | 71.34 |
| IRAK4 | 606883 | 100 | 100 | 98.98 | 91.73 |
| IRF2BP2 | 615332 | 100 | 98.97 | 87.56 | 64.19 |
| IRF3 | 603734 | 100 | 100 | 98.59 | 87.54 |
| IRF4 | 601900 | 100 | 99.21 | 92.80 | 90.58 |

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| IRF7 | 605047 | 100 | 98.70 | 96.44 | 84.98 |
| IRF8 | 601565 | 100 | 98.21 | 92.56 | 71.35 |
| IRF9 | 147574 | 100 | 100 | 99.44 | 91.97 |
| ISG15 | 147571 | 100 | 100 | 98.60 | 92.43 |
| ITCH | 606409 | 98.02 | 96.02 | 93.74 | 88.49 |
| ITGB2 | 600065 | 100 | 98.83 | 96.43 | 85.40 |
| ITK | 186973 | 100 | 100 | 99.73 | 97.05 |
| IVNS1ABP | 609209 | 100 | 100 | 100 | 100 |
| JAGN1 | 616012 | 100 | 100 | 100 | 100 |
| JAK1 | 147795 | 100 | 100 | 99.37 | 95.00 |
| JAK2 | 147796 | 100 | 99.39 | 97.94 | 93.19 |
| JAK3 | 600173 | 100 | 99.67 | 98.07 | 91.75 |
| KDM6A | 300128 | 100 | 98.66 | 94.85 | 78.84 |
| KMT2A | 159555 | 99.92 | 99.27 | 98.22 | 94.84 |
| KMT2D | 602113 | 100 | 100 | 99.75 | 95.33 |
| KRAS | 190070 | 100 | 100 | 100 | 100 |
| LACC1 | 613409 | 100 | 100 | 100 | 96.31 |
| LAMTOR2 | 610389 | 100 | 100 | 100 | 90.88 |
| LAT | 602354 | 100 | 100 | 97.06 | 77.64 |
| LCK | 153390 | 100 | 99.58 | 95.98 | 82.06 |
| LIG1 | 126391 | 100 | 98.76 | 94.18 | 78.45 |
| LIG4 | 601837 | 96.16 | 96.16 | 96.16 | 96.16 |
| LPIN2 | 605519 | 100 | 100 | 99.77 | 97.16 |
| LRBA | 606453 | 99.80 | 99.46 | 98.51 | 95.25 |
| LRRC8A | 608360 | 100 | 100 | 100 | 98.91 |
| LYST | 606897 | 100.00 | 99.81 | 98.90 | 94.04 |
| MAGT1 | 300715 | 99.69 | 93.27 | 90.85 | 83.63 |
| MAL | 188860 | 100 | 100 | 100 | 95.27 |
| MALT1 | 604860 | 95.57 | 91.25 | 90.18 | 88.28 |
| MAN2B1 | 609458 | 100 | 99.81 | 96.81 | 85.87 |
| MANBA | 609489 | 100 | 99.97 | 99.66 | 97.72 |
| MAP3K14 | 604655 | 100 | 98.96 | 95.36 | 90.52 |
| MASP2 | 605102 | 100 | 100 | 96.79 | 88.24 |
| MBL2 | 154545 | 100 | 100 | 100 | 96.97 |
| MC2R | 607397 | 100 | 100 | 100 | 100 |
| MCM4 | 602638 | 100 | 98.62 | 96.55 | 92.57 |
| MEFV | 608107 | 100 | 100 | 99.10 | 92.94 |
| MOGS | 601336 | 100 | 100 | 99.90 | 97.07 |
| MRE11 | 600814 | 100 | 100 | 99.52 | 95.09 |
| MRTFA | 606078 | 92.22 | 90.37 | 86.52 | 72.07 |
| MS4A1 | 112210 | 100 | 97.38 | 93.47 | 71.02 |
| MSN | 309845 | 100 | 100 | 99.35 | 91.26 |
| MTHFD1 | 172460 | 100 | 100 | 99.48 | 94.30 |
| MVK | 251170 | 100 | 100 | 100 | 96.47 |

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| MYD88 | 602170 | 100 | 100 | 100 | 97.12 |
| MYSM1 | 612176 | 99.69 | 96.78 | 93.08 | 89.62 |
| NBAS | 608025 | 100 | 100 | 99.61 | 95.26 |
| NBN | 602667 | 100 | 100 | 100 | 96.80 |
| NCF1 | 608512 | 58.99 | 51.83 | 39.42 | 24.06 |
| NCF2 | 608515 | 100 | 100 | 100 | 98.11 |
| NCF4 | 601488 | 100 | 100 | 98.40 | 83.32 |
| NCKAP1L | 141180 | 100 | 99.85 | 97.98 | 92.55 |
| NCSTN | 605254 | 100 | 100 | 100 | 98.28 |
| NFAT5 | 604708 | 100 | 99.56 | 98.05 | 94.16 |
| NFE2L2 | 600492 | 100 | 100 | 99.61 | 96.61 |
| NFKB1 | 164011 | 100 | 100 | 98.90 | 93.54 |
| NFKB2 | 164012 | 100 | 99.68 | 98.22 | 87.82 |
| NFKBIA | 164008 | 100 | 100 | 100 | 93.18 |
| NHEJ1 | 611290 | 100 | 100 | 100 | 96.58 |
| NHP2 | 606470 | 100 | 100 | 100 | 100 |
| NLRC4 | 606831 | 100 | 100 | 100 | 99.81 |
| NLRP1 | 606636 | 100 | 100 | 99.71 | 96.84 |
| NLRP12 | 609648 | 100 | 100 | 99.26 | 95.07 |
| NLRP3 | 606416 | 100 | 100 | 99.92 | 98.10 |
| NOD2 | 605956 | 100 | 100 | 99.78 | 96.90 |
| NOP10 | 606471 | 100 | 100 | 100 | 100 |
| NRAS | 164790 | 100 | 100 | 100 | 99.07 |
| NSMCE3 | 608243 | 100 | 100 | 100 | 96.08 |
| OAS1 | 164350 | 100 | 97.94 | 95.89 | 87.63 |
| ORAI1 | 610277 | 99.37 | 94.42 | 91.99 | 88.36 |
| OSTM1 | 607649 | 100 | 91.53 | 77.50 | 62.48 |
| OTULIN | 615712 | 91.89 | 85.62 | 85.62 | 80.64 |
| PARN | 604212 | 100 | 100 | 100 | 94.26 |
| PAX5 | 167414 | 100 | 100 | 100 | 97.18 |
| PBX1 | 176310 | 100 | 98.10 | 90.92 | 78.16 |
| PCCA | 232000 | 99.83 | 96.72 | 95.31 | 89.85 |
| PCCB | 232050 | 100 | 100 | 99.17 | 92.49 |
| PEPD | 613230 | 100 | 100 | 98.29 | 89.21 |
| PGM3 | 172100 | 100 | 100 | 100 | 98.86 |
| PIGA | 311770 | 100 | 100 | 96.12 | 90 |
| PIK3CD | 602839 | 100 | 99.94 | 99.44 | 97.51 |
| PIK3R1 | 171833 | 100 | 100 | 99.70 | 95.35 |
| PLCG2 | 600220 | 100 | 100 | 99.33 | 93.91 |
| PLEKHM1 | 611466 | 97.99 | 97.34 | 95.00 | 80.76 |
| PLG | 173350 | 100 | 100 | 94.67 | 85.86 |
| PMM2 | 601785 | 100 | 100 | 100 | 95.16 |
| PNP | 164050 | 100 | 100 | 100 | 99.19 |
| POLA1 | 312040 | 100 | 98.83 | 92.93 | 75.88 |

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|---------------------------|--|------------------|----------------|----------------|----------------|
| POLE2 | 602670 | 100 | 96.99 | 90.72 | 80.52 |
| POMP | 613386 | 99.08 | 85.08 | 82.78 | 80.85 |
| POT1 | 606478 | 100 | 100 | 98.75 | 92.23 |
| POU2AF1 | 601206 | 100 | 98.50 | 93.55 | 71.08 |
| PRF1 | 170280 | 100 | 100 | 100 | 100 |
| PRKCD | 176977 | 97.63 | 97.63 | 97.63 | 96.24 |
| PRKDC | 600899 | 99.75 | 98.97 | 98.40 | 94.68 |
| PRPS1 | 311850 | 100 | 100 | 100 | 92.79 |
| PSENFEN | 607632 | 100 | 100 | 98.62 | 89.26 |
| PSMA3 | 176843 | 100 | 100 | 100 | 93.20 |
| PSMB4 | 602177 | 100 | 100 | 98.66 | 92.97 |
| PSMB8 | 177046 | 100 | 100 | 100 | 99.26 |
| PSMB9 | 177045 | 100 | 100 | 98.84 | 89.70 |
| PSMG2 | 609702 | 100 | 100 | 100 | 97.64 |
| PSTPIP1 | 606347 | 100 | 100 | 99.89 | 88.43 |
| PTPN22 | 600716 | 98.20 | 98.20 | 98.17 | 96.87 |
| PTPRC | 151460 | 99.69 | 97.76 | 94.03 | 86.79 |
| RAB27A | 603868 | 100 | 100 | 100 | 99.54 |
| RAC2 | 602049 | 100 | 100 | 100 | 98.42 |
| RAG1 | 179615 | 100 | 100 | 100 | 100 |
| RAG2 | 179616 | 100 | 100 | 100 | 100 |
| RANBP2 | 601181 | 99.14 | 98.07 | 97.49 | 93.41 |
| RASGRP1 | 603962 | 100 | 100 | 100 | 98.41 |
| RASGRP2 | 605577 | 100 | 100 | 98.57 | 89.84 |
| RBCK1 | 610924 | 100 | 100 | 100 | 88.80 |
| RC3H1 | 609424 | 100 | 99.55 | 98.37 | 92.60 |
| RECQL4 | 603780 | 100 | 99.84 | 98.15 | 93.37 |
| RELB | 604758 | 100 | 99.04 | 95.32 | 75.87 |
| RFX5 | 601863 | 100 | 100 | 100 | 98.57 |
| RFXANK | 603200 | 100 | 100 | 98.24 | 85.32 |
| RFXAP | 601861 | 100 | 99.89 | 97.15 | 85.62 |
| RHOG | 179505 | 100 | 100 | 100 | 100 |
| RHOH | 602037 | 100 | 100 | 100 | 100 |
| RIPK1 | 603453 | 100 | 100 | 98.60 | 92.41 |
| RMRP | 157660 | No coverage data | | | |
| RNASEH2A | 606034 | 100 | 100 | 98.86 | 85.19 |
| RNASEH2B | 610326 | 100 | 91.35 | 91.35 | 91.35 |
| RNASEH2C | 610330 | 100 | 100 | 100 | 97.20 |
| RNF168 | 612688 | 100 | 100 | 100 | 99.13 |
| RNF31 | 612487 | 100 | 99.93 | 98.90 | 95.41 |
| RNU4ATAC | 601428 | No coverage data | | | |
| RORC | 602943 | 100 | 99.55 | 94.93 | 77.93 |
| RPSA | 150370 | 100 | 100 | 100 | 97.55 |
| RSPH9 | 612648 | 100 | 100 | 100 | 98.71 |

| HGNC approved gene symbol | OMIM gene ID (active link to omim.org) | % covered ≥10x | % covered ≥20x | % covered ≥30x | % covered ≥50x |
|---------------------------|--|----------------|----------------|----------------|----------------|
| RTEL1 | 608833 | 100 | 100 | 99.24 | 90.23 |
| SAMD9 | 610456 | 100 | 100 | 100 | 99.46 |
| SAMD9L | 611170 | 100 | 100 | 100 | 100 |
| SAMHD1 | 606754 | 100 | 100 | 100 | 99.98 |
| SBDS | 607444 | 100 | 100 | 100 | 100 |
| SEC61A1 | 609213 | 100 | 100 | 100 | 97.96 |
| SEMA3E | 608166 | 100 | 97.67 | 93.47 | 89.27 |
| SERAC1 | 614725 | 100 | 100 | 100 | 97.87 |
| SERPING1 | 606860 | 100 | 97.53 | 95.12 | 90.34 |
| SH2B3 | 605093 | 100 | 99.95 | 95.23 | 75.63 |
| SH2D1A | 300490 | 100 | 100 | 100 | 89.33 |
| SH3BP2 | 602104 | 91.15 | 91.15 | 91.15 | 89.92 |
| SH3KBP1 | 300374 | 100 | 98.45 | 95.71 | 84.43 |
| SKIV2L | 600478 | 100 | 100 | 98.56 | 92.86 |
| SLC29A3 | 612373 | 100 | 97.60 | 97.60 | 97.14 |
| SLC35A1 | 605634 | 100 | 97.23 | 96.30 | 94.46 |
| SLC35C1 | 605881 | 100 | 100 | 100 | 100 |
| SLC37A4 | 602671 | 99.87 | 99.87 | 99.10 | 93.11 |
| SLC39A4 | 607059 | 100 | 100 | 100 | 96.45 |
| SLC39A7 | 601416 | 100 | 100 | 100 | 98.22 |
| SLC46A1 | 611672 | 99.87 | 99.87 | 99.87 | 97.80 |
| SLC7A7 | 603593 | 100 | 100 | 100 | 97.31 |
| SMARCAL1 | 606622 | 100 | 100 | 99.62 | 95.10 |
| SMARCD2 | 601736 | 87.26 | 87.26 | 87.26 | 85.08 |
| SNX10 | 614780 | 100 | 100 | 100 | 97.57 |
| SOCS4 | 616337 | 100 | 100 | 99.37 | 94.81 |
| SP110 | 604457 | 100 | 100 | 99.26 | 91.22 |
| SPINK5 | 605010 | 100 | 97.65 | 92.50 | 79.16 |
| SPPL2A | 608238 | 97.69 | 94.44 | 89.27 | 84.19 |
| SRP54 | 604857 | 99.45 | 97.93 | 95.86 | 84.71 |
| SRP72 | 602122 | 100 | 100 | 100 | 98.70 |
| STAT1 | 600555 | 97.05 | 97.05 | 97.05 | 96.66 |
| STAT2 | 600556 | 100 | 100 | 100 | 98.08 |
| STAT3 | 102582 | 100 | 100 | 99.75 | 94.86 |
| STAT4 | 600558 | 99.93 | 97.04 | 96.28 | 94.05 |
| STAT5B | 604260 | 100 | 98.35 | 96.03 | 87.06 |
| STAT6 | 601512 | 100 | 99.49 | 95.86 | 80.87 |
| STIM1 | 605921 | 100 | 100 | 100 | 97.95 |
| STING1 | 612374 | 100 | 100 | 100 | 88.46 |
| STK4 | 604965 | 100 | 97.06 | 96.04 | 88.85 |
| STN1 | 613128 | 100 | 100 | 100 | 99.30 |
| STX11 | 605014 | 100 | 100 | 100 | 100 |
| STXBP2 | 601717 | 100 | 97.15 | 92.77 | 77.46 |
| TAP1 | 170260 | 100 | 99.79 | 97.84 | 86.65 |

| HGNC approved gene symbol | OMIM gene ID (active link to omim.org) | % covered $\geq 10x$ | % covered $\geq 20x$ | % covered $\geq 30x$ | % covered $\geq 50x$ |
|---------------------------|--|----------------------|----------------------|----------------------|----------------------|
| TAP2 | 170261 | 100 | 98.55 | 95.30 | 87.73 |
| TAPBP | 601962 | 100 | 98.61 | 94.64 | 81.20 |
| TAZ | 300394 | 100 | 100 | 98.38 | 90.52 |
| TBX1 | 602054 | 91.70 | 82.58 | 69.57 | 45.38 |
| TCF3 | 147141 | 100 | 99.57 | 92.74 | 71.53 |
| TCIRG1 | 604592 | 100 | 97.68 | 92.86 | 83.01 |
| TCN2 | 613441 | 100 | 100 | 99.90 | 95.24 |
| TERC | 602322 | No coverage data | | | |
| TERT | 187270 | 100 | 98.75 | 95.08 | 87.33 |
| TET2 | 612839 | 100 | 99.98 | 99.14 | 96.81 |
| TFRC | 190010 | 100 | 99.92 | 98.75 | 96.31 |
| TGFB1 | 190180 | 100 | 100 | 100 | 99.01 |
| THBD | 188040 | 100 | 100 | 100 | 95.64 |
| TICAM1 | 607601 | 100 | 100 | 98.03 | 84.97 |
| TINF2 | 604319 | 100 | 100 | 100 | 99.35 |
| TIRAP | 606252 | 100 | 100 | 100 | 100 |
| TLR3 | 603029 | 100 | 100 | 100 | 98.35 |
| TLR4 | 603030 | 100 | 100 | 100 | 100 |
| TLR7 | 300365 | 100 | 100 | 100 | 99.45 |
| TLR8 | 300366 | 100 | 100 | 100 | 99.72 |
| TMC6 | 605828 | 100 | 100 | 99.48 | 85.31 |
| TMC8 | 605829 | 100 | 99.46 | 92.51 | 65.36 |
| TNFAIP3 | 191163 | 100 | 100 | 99.01 | 96.82 |
| TNFRSF11A | 603499 | 90.20 | 90.20 | 87.25 | 80.86 |
| TNFRSF13B | 604907 | 100 | 100 | 98.77 | 90.35 |
| TNFRSF13C | 606269 | 91.34 | 66.50 | 60.13 | 51.23 |
| TNFRSF1A | 191190 | 100 | 100 | 100 | 92.91 |
| TNFRSF4 | 600315 | 100 | 98.97 | 87.13 | 47.32 |
| TNFRSF9 | 602250 | 100 | 100 | 97.57 | 84.03 |
| TNFSF11 | 602642 | 100 | 100 | 100 | 98.03 |
| TNFSF12 | 602695 | 94.70 | 86.53 | 71.70 | 55.36 |
| TOM1 | 604700 | 100 | 100 | 100 | 97.16 |
| TOP2B | 126431 | 99.78 | 98.74 | 95.63 | 85.85 |
| TPP2 | 190470 | 96.67 | 95.13 | 91.65 | 83.91 |
| TRAF3 | 601896 | 100 | 100 | 98.90 | 90.65 |
| TRAF3IP2 | 607043 | 100 | 100 | 100 | 99.35 |
| TREX1 | 606609 | 100 | 100 | 100 | 100 |
| TRIM22 | 606559 | 100 | 100 | 98.23 | 87.85 |
| TRNT1 | 612907 | 99.83 | 90.53 | 89.36 | 83.15 |
| TTC37 | 614589 | 100 | 100 | 99.48 | 97.36 |
| TTC7A | 609332 | 100 | 100 | 99.05 | 90.00 |
| TYK2 | 176941 | 100 | 100 | 99.70 | 94.95 |
| UBA1 | 314370 | 100 | 99.76 | 98.07 | 90.22 |
| UNC13D | 608897 | 100 | 99.85 | 98.15 | 88.87 |

| HGNC approved gene symbol | OMIM gene ID (active link to omim.org) | % covered $\geq 10x$ | % covered $\geq 20x$ | % covered $\geq 30x$ | % covered $\geq 50x$ |
|---------------------------|--|----------------------|----------------------|----------------------|----------------------|
| UNC93B1 | 608204 | 75.96 | 69.28 | 67.31 | 59.79 |
| UNG | 191525 | 100 | 100 | 100 | 88.65 |
| USB1 | 613276 | 100 | 96.58 | 86.74 | 75.56 |
| USP18 | 607057 | 95.21 | 95.21 | 95.21 | 90.35 |
| VAV1 | 164875 | 100 | 99.79 | 98 | 92.29 |
| VPS13B | 607817 | 99.15 | 98.02 | 95.90 | 88.67 |
| VPS45 | 610035 | 100 | 100 | 99.88 | 96.32 |
| WAS | 300392 | 100 | 100 | 98.42 | 83.59 |
| WDR1 | 604734 | 100 | 99.83 | 98.30 | 97.17 |
| WIPF1 | 602357 | 100 | 98.97 | 96.18 | 77.02 |
| WRAP53 | 612661 | 100 | 100 | 100 | 95.04 |
| XIAP | 300079 | 100 | 100 | 95.53 | 84.45 |
| ZAP70 | 176947 | 100 | 99.67 | 97.76 | 91.34 |
| ZBTB24 | 614064 | 100 | 100 | 100 | 98.96 |
| ZNF341 | 618269 | 99.21 | 98.08 | 92.42 | 83.42 |

- OMIM release used: 18-2-2021

- The statistics above are based on a set of 100 samples

- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence ($\pm 10bp$ flanking introns) that is covered at least 10x, 20x, 30x or 50x