

# Whole Exome Sequencing

## Gene package Skeletal Dysplasia, Version 3, 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 | % covered ≥10x | % covered ≥20x | % covered ≥30x | % covered ≥50x |
|---------------------------|--------------|----------------|----------------|----------------|----------------|
| ABCC9                     | 601439       | 100            | 99.86          | 98.89          | 95.39          |
| ACAN                      | 155760       | 92.87          | 92.06          | 91.16          | 87.95          |
| ACP5                      | 171640       | 100            | 100            | 100            | 100            |
| ACTB                      | 102630       | 100            | 100            | 100            | 98.33          |
| ACVR1                     | 102576       | 100            | 100            | 100            | 96.28          |
| ADAMTS10                  | 608990       | 100            | 100            | 99.33          | 90.42          |
| ADAMTS17                  | 607511       | 95.60          | 90.58          | 86.35          | 81.11          |
| ADAMTSL2                  | 612277       | 44.82          | 42.41          | 39.31          | 32.66          |
| AGA                       | 613228       | 100            | 100            | 98.65          | 89.74          |
| AGPS                      | 603051       | 95.91          | 95.28          | 93.05          | 87.66          |
| ALG12                     | 607144       | 100            | 100            | 100            | 99.36          |
| ALG3                      | 608750       | 100            | 100            | 98.80          | 90.09          |
| ALG9                      | 606941       | 99.91          | 99.68          | 97.21          | 87.85          |
| ALMS1                     | 606844       | 99.78          | 99.08          | 97.76          | 93.88          |
| ALPL                      | 171760       | 100            | 100            | 100            | 98.94          |
| AMER1                     | 300647       | 100            | 100            | 100            | 100            |
| ANKH                      | 605145       | 100            | 100            | 100            | 94.46          |
| ANKRD11                   | 611192       | 100            | 98.06          | 96.47          | 92.67          |
| ANO5                      | 608662       | 100            | 99.31          | 96.43          | 89.27          |
| ARSB                      | 611542       | 100            | 99.83          | 97.73          | 84.48          |
| ARSL                      | 300180       | 100            | 100            | 99.44          | 92.19          |
| B3GALT6                   | 615291       | 78.50          | 74.68          | 72.00          | 67.78          |
| B3GAT3                    | 606374       | 100            | 100            | 100            | 89.82          |
| B4GALT7                   | 604327       | 99.95          | 93.64          | 93.28          | 84.20          |

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|---------------------------|--------------|----------------|----------------|----------------|----------------|
| BMP1                      | 112264       | 100            | 100            | 98.99          | 91.87          |
| BMPER                     | 608699       | 100            | 99.15          | 97.54          | 92.97          |
| BMPR1B                    | 603248       | 100            | 100            | 97.99          | 91.96          |
| BRAF                      | 164757       | 100            | 99.25          | 98.19          | 92.07          |
| BTK                       | 300300       | 100            | 100            | 99.69          | 93.31          |
| CA2                       | 611492       | 100            | 100            | 100            | 97.88          |
| CANT1                     | 613165       | 100            | 100            | 100            | 98.85          |
| CBL                       | 165360       | 100            | 100            | 100            | 97.75          |
| CCDC8                     | 614145       | 100            | 100            | 99.63          | 96.18          |
| CCN6                      | 603400       | 100            | 100            | 97.72          | 89.76          |
| CDC45                     | 603465       | 100            | 100            | 100            | 96.18          |
| CDC6                      | 602627       | 100            | 100            | 100            | 97.84          |
| CDKN1C                    | 600856       | 84.21          | 69.74          | 58.81          | 48.73          |
| CDT1                      | 605525       | 99.65          | 93.55          | 87.76          | 73.86          |
| CEP120                    | 613446       | 100            | 100            | 99.96          | 95.96          |
| CFAP410                   | 603191       | 100            | 100            | 97.85          | 76.65          |
| CHST3                     | 603799       | 100            | 100            | 100            | 97.12          |
| CLCN5                     | 300008       | 100            | 99.76          | 98.36          | 88.33          |
| CLCN7                     | 602727       | 100            | 100            | 99.30          | 89.11          |
| COG1                      | 606973       | 100            | 100            | 99.80          | 95.47          |
| COL10A1                   | 120110       | 100            | 100            | 99.13          | 96.49          |
| COL11A1                   | 120280       | 100            | 98.68          | 95.94          | 84.52          |
| COL11A2                   | 120290       | 100            | 99.07          | 94.23          | 80.93          |
| COL1A1                    | 120150       | 100            | 99.67          | 96.63          | 82.83          |
| COL1A2                    | 120160       | 99.88          | 97.88          | 94.75          | 82.39          |
| COL2A1                    | 120140       | 100            | 99.83          | 98.57          | 91.36          |
| COL9A1                    | 120210       | 100            | 99.65          | 96.88          | 80.71          |
| COL9A2                    | 120260       | 100            | 95.05          | 86.28          | 57.61          |
| COL9A3                    | 120270       | 100            | 99.63          | 94.99          | 74.85          |
| COLEC11                   | 612502       | 100            | 100            | 97.79          | 89.50          |
| COMP                      | 600310       | 98.40          | 92.72          | 92.55          | 83.95          |
| CREB3L1                   | 616215       | 99.94          | 97.33          | 92.43          | 77.77          |
| CREBBP                    | 600140       | 99.64          | 98.59          | 96.38          | 88.55          |
| CRTAP                     | 605497       | 100            | 100            | 100            | 95.83          |
| CSGALNACT1                | 616615       | 100            | 100            | 100            | 100            |
| CTSA                      | 613111       | 100            | 99.89          | 99.16          | 90.94          |
| CTSK                      | 601105       | 100            | 100            | 100            | 98.27          |
| CUL7                      | 609577       | 100            | 100            | 98.67          | 96.19          |
| CYP26B1                   | 605207       | 100            | 100            | 99.58          | 94.02          |
| CYP27B1                   | 609506       | 100            | 100            | 99.82          | 89.44          |
| DDR2                      | 191311       | 100            | 100            | 100            | 99.08          |
| DHCR24                    | 606418       | 100            | 100            | 100            | 96.21          |
| DLL3                      | 602768       | 100            | 98.31          | 88.08          | 62.26          |
| DLX3                      | 600525       | 100            | 100            | 98.37          | 86.75          |

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|---------------------------|--------------|----------------|----------------|----------------|----------------|
| DMP1                      | 600980       | 100            | 100            | 100            | 93.99          |
| DONSON                    | 611428       | 96.97          | 93.52          | 88.33          | 75.58          |
| DPM1                      | 603503       | 100            | 100            | 100            | 97.03          |
| DVL1                      | 601365       | 99.27          | 92.68          | 88.91          | 74.44          |
| DYM                       | 607461       | 100            | 100            | 99.64          | 95.06          |
| DYNC2H1                   | 603297       | 99.06          | 97.98          | 95.50          | 87.54          |
| EBP                       | 300205       | 100            | 100            | 100            | 97.34          |
| EIF2AK3                   | 604032       | 99.92          | 97.00          | 93.56          | 86.97          |
| ENPP1                     | 173335       | 92.81          | 91.15          | 88.95          | 85.86          |
| EVC                       | 604831       | 94.29          | 93.49          | 91.71          | 82.76          |
| EVC2                      | 607261       | 100            | 99.70          | 98.53          | 93.34          |
| EXT1                      | 608177       | 100            | 100            | 100            | 97.60          |
| EXT2                      | 608210       | 100            | 100            | 100            | 98.86          |
| EXTL3                     | 605744       | 100            | 100            | 100            | 100            |
| FAM111A                   | 615292       | 100            | 100            | 99.25          | 97.17          |
| FAM20C                    | 611061       | 100            | 96.88          | 92.09          | 73.00          |
| FBN1                      | 134797       | 100            | 100            | 99.70          | 98.30          |
| FERMT3                    | 607901       | 100            | 98.11          | 93.07          | 84.17          |
| FGD1                      | 300546       | 100            | 98.23          | 95.76          | 87.62          |
| FGF23                     | 605380       | 100            | 100            | 100            | 100            |
| FGF8                      | 600483       | 93.90          | 88.15          | 87.21          | 62.09          |
| FGF9                      | 600921       | 100            | 100            | 99.20          | 91.23          |
| FGFR1                     | 136350       | 100            | 100            | 99.61          | 96.92          |
| FGFR2                     | 176943       | 100            | 100            | 99.54          | 97.18          |
| FGFR3                     | 134934       | 100            | 99.62          | 98.10          | 84.77          |
| FIG4                      | 609390       | 100            | 100            | 100            | 98.04          |
| FKBP10                    | 607063       | 100            | 99.61          | 96.89          | 88.44          |
| FLNA                      | 300017       | 100            | 99.43          | 98.63          | 95.60          |
| FLNB                      | 603381       | 100            | 100            | 99.61          | 95.58          |
| FN1                       | 135600       | 100            | 100            | 99.80          | 96.96          |
| FUCA1                     | 612280       | 100            | 100            | 100            | 98.59          |
| FZD2                      | 600667       | 99.56          | 97.55          | 96.15          | 92.07          |
| GALNS                     | 612222       | 100            | 100            | 98.31          | 83.58          |
| GALNT3                    | 601756       | 100            | 100            | 100            | 97.45          |
| GDF3                      | 606522       | 100            | 100            | 100            | 96.02          |
| GDF5                      | 601146       | 100            | 100            | 100            | 98.41          |
| GDF6                      | 601147       | 100            | 100            | 97.33          | 69.64          |
| GH1                       | 139250       | 100            | 100            | 100            | 100            |
| GHR                       | 600946       | 100            | 100            | 99.06          | 97.52          |
| GHRH                      | 139190       | 100            | 100            | 98.51          | 74.50          |
| GHRHR                     | 139191       | 100            | 98.01          | 93.66          | 80.41          |
| GHSR                      | 601898       | 100            | 100            | 100            | 100            |
| GJA1                      | 121014       | 100            | 100            | 100            | 100            |
| GLB1                      | 611458       | 100            | 100            | 99.78          | 97.42          |

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| GLI2                      | 165230       | 99.97          | 98.54          | 95.29          | 87.06          |
| GLI3                      | 165240       | 100            | 100            | 100            | 98.10          |
| GMNN                      | 602842       | 100            | 95.11          | 85.07          | 59.71          |
| GNAS                      | 139320       | 100            | 100            | 100            | 97.95          |
| GNPAT                     | 602744       | 100            | 100            | 98.71          | 94.62          |
| GNPTAB                    | 607840       | 100            | 99.92          | 99.10          | 96.36          |
| GNPTG                     | 607838       | 100            | 97.22          | 94.10          | 93.66          |
| GNS                       | 607664       | 100            | 97.75          | 95.87          | 90.42          |
| GORAB                     | 607983       | 100            | 99.27          | 94.32          | 79.22          |
| GPC6                      | 604404       | 100            | 100            | 99.95          | 96.72          |
| GPX4                      | 138322       | 100            | 96.14          | 92.99          | 88.42          |
| GUSB                      | 611499       | 100            | 100            | 100            | 96.42          |
| HDAC4                     | 605314       | 99.47          | 96.01          | 94.09          | 81.03          |
| HES7                      | 608059       | 90.84          | 70.39          | 59.55          | 33.64          |
| HESX1                     | 601802       | 100            | 96.69          | 91.50          | 78.82          |
| HGSNAT                    | 610453       | 94.18          | 93.19          | 91.08          | 81.62          |
| HMGA2                     | 600698       | 62.38          | 58.83          | 45.68          | 25.14          |
| HOXA13                    | 142959       | 83.43          | 77.66          | 73.38          | 66.03          |
| HPGD                      | 601688       | 100            | 100            | 99.15          | 94.72          |
| HRAS                      | 190020       | 100            | 100            | 100            | 100            |
| HSPA9                     | 600548       | 100            | 99.83          | 98.57          | 95.75          |
| HSPG2                     | 142461       | 99.45          | 98.73          | 96.55          | 87.82          |
| HYLS1                     | 610693       | 100            | 100            | 100            | 96.29          |
| IDH2                      | 147650       | 100            | 97.14          | 91.43          | 90.64          |
| IDS                       | 300823       | 100            | 99.07          | 94.84          | 84.56          |
| IDUA                      | 252800       | 98.66          | 89.77          | 84.08          | 70.92          |
| IFITM5                    | 614757       | 100            | 100            | 100            | 100            |
| IFT122                    | 606045       | 100            | 99.83          | 98.59          | 91.64          |
| IFT140                    | 614620       | 100            | 100            | 98.91          | 92.50          |
| IFT172                    | 607386       | 100            | 100            | 99.30          | 96.91          |
| IFT43                     | 614068       | 100            | 100            | 100            | 97.33          |
| IFT80                     | 611177       | 100            | 99.93          | 97.09          | 85.61          |
| IGF1                      | 147440       | 100            | 100            | 100            | 89.32          |
| IGF1R                     | 147370       | 100            | 100            | 99.00          | 95.39          |
| IGF2                      | 147470       | 100            | 100            | 100            | 93.97          |
| IGFALS                    | 601489       | 100            | 100            | 100            | 95.89          |
| IGFBP3                    | 146732       | 100            | 100            | 99.59          | 79.51          |
| IGSF1                     | 300137       | 100            | 100            | 100            | 96.40          |
| IHH                       | 600726       | 100            | 99.81          | 97.25          | 92.85          |
| IKBKB                     | 603258       | 100            | 99.81          | 98.47          | 90.57          |
| IKBKG                     | 300248       | 33.32          | 25.95          | 25.95          | 22.46          |
| IL2RG                     | 308380       | 100            | 100            | 99.61          | 90.29          |
| IMPAD1                    | 614010       | 100            | 100            | 100            | 97.71          |
| INPPL1                    | 600829       | 98.64          | 95.34          | 94.44          | 85.87          |

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|---------------------------|--------------|----------------|----------------|----------------|----------------|
| KIAA0753                  | 617112       | 100            | 99.88          | 98.91          | 95.88          |
| KIF22                     | 603213       | 100            | 100            | 98.64          | 93.03          |
| KIF7                      | 611254       | 99.65          | 96.04          | 90.61          | 78.96          |
| KMT2A                     | 159555       | 99.92          | 99.27          | 98.22          | 94.84          |
| KRAS                      | 190070       | 100            | 100            | 100            | 100            |
| LBR                       | 600024       | 100            | 100            | 97.36          | 90.52          |
| LEMD3                     | 607844       | 100            | 99.50          | 96.58          | 83.81          |
| LFNG                      | 602576       | 100            | 100            | 100            | 89.62          |
| LHX3                      | 600577       | 100            | 99.77          | 96.91          | 80.09          |
| LHX4                      | 602146       | 100            | 100            | 98.53          | 89.57          |
| LIFR                      | 151443       | 100            | 98.34          | 93.34          | 85.44          |
| LMX1B                     | 602575       | 100            | 96.26          | 91.26          | 85.12          |
| LONP1                     | 605490       | 100            | 99.61          | 97.64          | 90.32          |
| LRP4                      | 604270       | 100            | 100            | 99.51          | 96.33          |
| LRP5                      | 603506       | 97.91          | 97.83          | 96.64          | 91.64          |
| LRRK1                     | 610986       | 99.76          | 98.60          | 96.52          | 90.63          |
| LTBP2                     | 602091       | 99.95          | 98.66          | 94.89          | 84.52          |
| LTBP3                     | 602090       | 99.31          | 96.97          | 90.24          | 73.36          |
| LZTR1                     | 600574       | 100            | 100            | 100            | 96.12          |
| MAN2B1                    | 609458       | 100            | 99.81          | 96.81          | 85.87          |
| MANBA                     | 609489       | 100            | 99.97          | 99.66          | 97.72          |
| MAP2K1                    | 176872       | 100            | 100            | 99.21          | 93.07          |
| MAP2K2                    | 601263       | 100            | 99.89          | 96.23          | 84.15          |
| MAP3K7                    | 602614       | 100            | 99.51          | 98.68          | 95.06          |
| MATN3                     | 602109       | 91.47          | 85.78          | 84.98          | 84.98          |
| MBTPS2                    | 300294       | 100            | 97.61          | 93.92          | 88.10          |
| MEOX1                     | 600147       | 100            | 100            | 100            | 78.41          |
| MESP2                     | 605195       | 100            | 98.01          | 95.69          | 83.31          |
| MGP                       | 154870       | 100            | 91.01          | 89.05          | 87.60          |
| MMP13                     | 600108       | 100            | 100            | 100            | 98.05          |
| MMP14                     | 600754       | 100            | 100            | 99.87          | 94.17          |
| MMP2                      | 120360       | 100            | 99.87          | 98.26          | 87.92          |
| MMP9                      | 120361       | 100            | 100            | 99.45          | 89.37          |
| MTAP                      | 156540       | 100            | 100            | 100            | 98.81          |
| MYH3                      | 160720       | 100            | 99.49          | 98.48          | 94.31          |
| MYO18B                    | 607295       | 100            | 99.63          | 97.64          | 88.11          |
| NAGLU                     | 609701       | 100            | 94.64          | 90.80          | 83.50          |
| NANS                      | 605202       | 100            | 100            | 99.42          | 92.31          |
| NBAS                      | 608025       | 100            | 100            | 99.61          | 95.26          |
| NEK1                      | 604588       | 100            | 98.60          | 97.14          | 89.44          |
| NEK9                      | 609798       | 100            | 99.94          | 98.34          | 93.00          |
| NEU1                      | 608272       | 100            | 100            | 100            | 99.05          |
| NIN                       | 608684       | 100            | 99.86          | 98.83          | 95.18          |
| NIPBL                     | 608667       | 100            | 99.77          | 98.78          | 95.02          |

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| NKX3-2                    | 602183       | 100            | 100            | 96.54          | 85.56          |
| NOTCH2                    | 600275       | 99.78          | 98.49          | 97.37          | 93.59          |
| NPPA                      | 108780       | 100            | 100            | 100            | 97.95          |
| NPPB                      | 600295       | 100            | 100            | 96.32          | 66.77          |
| NPPC                      | 600296       | 100            | 100            | 93.90          | 65.19          |
| NPR1                      | 108960       | 100            | 98.68          | 93.40          | 82.71          |
| NPR2                      | 108961       | 100            | 100            | 100            | 98.18          |
| NPR3                      | 108962       | 100            | 100            | 100            | 98.59          |
| NRAS                      | 164790       | 100            | 100            | 100            | 99.07          |
| OBSL1                     | 610991       | 100            | 100            | 99.10          | 94.20          |
| OFD1                      | 300170       | 100            | 98.67          | 94.05          | 75.14          |
| ORC1                      | 601902       | 100            | 100            | 100            | 97.19          |
| ORC4                      | 603056       | 100            | 100            | 99.62          | 89.92          |
| ORC6                      | 607213       | 100            | 100            | 99.11          | 84.65          |
| OSTM1                     | 607649       | 100            | 91.53          | 77.50          | 62.48          |
| OTX2                      | 600037       | 100            | 100            | 100            | 100            |
| P3H1                      | 610339       | 100            | 100            | 100            | 99.65          |
| P4HB                      | 176790       | 100            | 99.43          | 96.19          | 83.57          |
| PAM16                     | 614336       | 100            | 100            | 98.53          | 93.47          |
| PAPPA                     | 176385       | 95.44          | 94.16          | 91.93          | 89.37          |
| PAPPA2                    | No ID        | 100            | 100            | 99.49          | 96.37          |
| PAPSS2                    | 603005       | 100            | 100            | 99.57          | 90.27          |
| PCNT                      | 605925       | 100            | 99.35          | 96.70          | 90.36          |
| PCYT1A                    | 123695       | 100            | 100            | 100            | 90.68          |
| PDE4D                     | 600129       | 100            | 100            | 100            | 97.21          |
| PEX5                      | 600414       | 100            | 100            | 99.65          | 93.11          |
| PEX7                      | 601757       | 100            | 100            | 96.83          | 87.17          |
| PHEX                      | 300550       | 100            | 100            | 99.55          | 87.29          |
| PHGDH                     | 606879       | 100            | 100            | 99.35          | 93.12          |
| PHYH                      | 602026       | 99.89          | 89.33          | 89.33          | 76.91          |
| PIK3CA                    | 171834       | 100            | 100            | 99.94          | 97.86          |
| PIK3R1                    | 171833       | 100            | 100            | 99.70          | 95.35          |
| PIK3R3                    | 606076       | 100            | 100            | 100            | 95.25          |
| PITX1                     | 602149       | 99.70          | 93.71          | 85.18          | 73.05          |
| PITX2                     | 601542       | 100            | 96.68          | 92.15          | 80.99          |
| PLEKHM1                   | 611466       | 97.99          | 97.34          | 95.00          | 80.76          |
| PLK4                      | 605031       | 100            | 100            | 99.26          | 95.79          |
| PLOD2                     | 601865       | 96.28          | 91.42          | 88.74          | 84.89          |
| PLS3                      | 300131       | 100            | 100            | 99.36          | 88.95          |
| POC1A                     | 614783       | 100            | 100            | 100            | 97.36          |
| POP1                      | 602486       | 100            | 100            | 99.78          | 94.74          |
| POR                       | 124015       | 100            | 100            | 100            | 98.25          |
| POU1F1                    | 173110       | 99.40          | 91.84          | 90.03          | 83.84          |
| PPIB                      | 123841       | 100            | 100            | 100            | 99.73          |

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| PRKAR1A                   | 188830       | 100              | 100            | 100            | 98.92          |
| PROKR2                    | 607123       | 100              | 100            | 100            | 100            |
| PROP1                     | 601538       | 100              | 100            | 94.65          | 66.46          |
| PSAT1                     | 610936       | 100              | 100            | 98.22          | 89.15          |
| PTDSS1                    | 612792       | 100              | 100            | 100            | 97.08          |
| PTH1R                     | 168468       | 100              | 100            | 99.93          | 93.25          |
| PTPN11                    | 176876       | 98.37            | 98.37          | 98.37          | 98.28          |
| RAB33B                    | 605950       | 100              | 100            | 100            | 100            |
| RAF1                      | 164760       | 100              | 100            | 97.97          | 93.07          |
| RASGRP2                   | 605577       | 100              | 100            | 98.57          | 89.84          |
| RBM8A                     | 605313       | 100              | 100            | 100            | 92.60          |
| RBPJ                      | 147183       | 100              | 98.60          | 97.01          | 93.14          |
| RIPPLY2                   | 609891       | 100              | 100            | 98.60          | 81.14          |
| RIT1                      | 609591       | 100              | 100            | 100            | 100            |
| RMRP                      | 157660       | No coverage data |                |                |                |
| RNU4ATAC                  | 601428       | No coverage data |                |                |                |
| ROR2                      | 602337       | 100              | 99.58          | 97.96          | 92.62          |
| RPGRIP1L                  | 610937       | 99.01            | 97.45          | 94.84          | 90.66          |
| RPL10                     | 312173       | 100              | 100            | 100            | 100            |
| RSPRY1                    | 616585       | 100              | 100            | 100            | 99.73          |
| RUNX2                     | 600211       | 100              | 100            | 100            | 99.07          |
| SBDS                      | 607444       | 100              | 100            | 100            | 100            |
| SCARF2                    | 613619       | 96.90            | 88.73          | 76.61          | 60.02          |
| SCUBE3                    | 614708       | 100              | 99.75          | 97.78          | 96.02          |
| SEC24D                    | 607186       | 100              | 99.76          | 98.32          | 91.51          |
| SERPINF1                  | 172860       | 100              | 100            | 100            | 96.90          |
| SERPINH1                  | 600943       | 100              | 100            | 100            | 100            |
| SGSH                      | 605270       | 93.52            | 93.52          | 93.52          | 92.20          |
| SH3PXD2B                  | 613293       | 100              | 98.68          | 96.32          | 88.84          |
| SHOC2                     | 602775       | 100              | 100            | 100            | 97.82          |
| SHOX                      | 312865       | 100              | 100            | 96.47          | 77.31          |
| SHOX2                     | 602504       | 100              | 99.24          | 93.54          | 86.08          |
| SLC10A7                   | 611459       | 100              | 100            | 94.54          | 65.86          |
| SLC17A5                   | 604322       | 100              | 100            | 100            | 95.37          |
| SLC25A24                  | 608744       | 100              | 100            | 98.56          | 89.42          |
| SLC26A2                   | 606718       | 100              | 100            | 100            | 100            |
| SLC29A3                   | 612373       | 100              | 97.60          | 97.60          | 97.14          |
| SLC34A3                   | 609826       | 100              | 99.36          | 97.79          | 88.88          |
| SLC35D1                   | 610804       | 100              | 99.00          | 95.59          | 82.41          |
| SLC39A13                  | 608735       | 100              | 100            | 100            | 93.46          |
| SLCO2A1                   | 601460       | 100              | 99.59          | 97.15          | 81.05          |
| SLCO5A1                   | 613543       | 100              | 99.36          | 98.02          | 93.14          |
| SMAD4                     | 600993       | 100              | 100            | 100            | 98.00          |
| SMARCAL1                  | 606622       | 100              | 100            | 99.62          | 95.10          |

| HGNC approved gene symbol | OMIM gene ID | % covered ≥10x | % covered ≥20x | % covered ≥30x | % covered ≥50x |
|---------------------------|--------------|----------------|----------------|----------------|----------------|
| SNRPB                     | 182282       | 100            | 100            | 96.92          | 80.06          |
| SNX10                     | 614780       | 100            | 100            | 100            | 97.57          |
| SOS1                      | 182530       | 100            | 97.71          | 97.35          | 91.97          |
| SOS2                      | 601247       | 99.08          | 97.55          | 95.49          | 87.28          |
| SOST                      | 605740       | 100            | 100            | 100            | 100            |
| SOX2                      | 184429       | 100            | 100            | 98.76          | 91.40          |
| SOX3                      | 313430       | 100            | 99.89          | 95.47          | 76.99          |
| SOX9                      | 608160       | 100            | 97.67          | 94.80          | 84.47          |
| SP7                       | 606633       | 100            | 100            | 100            | 90.59          |
| SPARC                     | 182120       | 100            | 100            | 99.83          | 92.02          |
| SPINK5                    | 605010       | 100            | 97.65          | 92.50          | 79.16          |
| SPR                       | 182125       | 100            | 100            | 95.49          | 79.89          |
| SRCAP                     | 611421       | 100            | 99.97          | 99.02          | 94.52          |
| STAT3                     | 102582       | 100            | 100            | 99.75          | 94.86          |
| STAT5B                    | 604260       | 100            | 98.35          | 96.03          | 87.06          |
| SULF1                     | 610012       | 100            | 99.67          | 97.83          | 89.14          |
| SUMF1                     | 607939       | 100            | 100            | 100            | 100            |
| TAPT1                     | 612758       | 95.58          | 88.29          | 81.93          | 74.76          |
| TBCE                      | 604934       | 100            | 99.35          | 98.46          | 92.50          |
| TBX15                     | 604127       | 100            | 100            | 98.98          | 96.44          |
| TBX4                      | 601719       | 100            | 96.41          | 92.80          | 82.15          |
| TBX6                      | 602427       | 100            | 100            | 100            | 83.58          |
| TBXAS1                    | 274180       | 100            | 100            | 100            | 100            |
| TCIRG1                    | 604592       | 100            | 97.68          | 92.86          | 83.01          |
| TCTEX1D2                  | 617353       | 100            | 100            | 100            | 100            |
| TCTN2                     | 613846       | 100            | 100            | 99.08          | 93.90          |
| TCTN3                     | 613847       | 100            | 100            | 100            | 95.60          |
| TGFB1                     | 190180       | 100            | 100            | 100            | 99.01          |
| TMEM165                   | 614726       | 100            | 100            | 98.40          | 89.74          |
| TMEM216                   | 613277       | 100            | 100            | 100            | 96.69          |
| TMEM231                   | 614949       | 100            | 100            | 99.19          | 94.38          |
| TMEM38B                   | 611236       | 100            | 100            | 100            | 98.84          |
| TNFRSF11A                 | 603499       | 90.20          | 90.20          | 87.25          | 80.86          |
| TNFRSF11B                 | 602643       | 100            | 100            | 100            | 100            |
| TNFSF11                   | 602642       | 100            | 100            | 100            | 98.03          |
| TRAPPC2                   | 300202       | 100            | 100            | 96.70          | 64.07          |
| TRIP11                    | 604505       | 99.97          | 99.06          | 96.19          | 88.73          |
| TRPS1                     | 604386       | 100            | 100            | 100            | 100            |
| TRPV4                     | 605427       | 100            | 100            | 99.59          | 93.70          |
| TTC21B                    | 612014       | 100            | 100            | 100            | 98.62          |
| VDR                       | 601769       | 100            | 95.13          | 90.07          | 76.31          |
| WDR19                     | 608151       | 100            | 99.81          | 98.99          | 94.93          |
| WDR34                     | 613363       | 100            | 99.78          | 96.90          | 76.51          |
| WDR35                     | 613602       | 99.99          | 99.40          | 97.68          | 94.88          |



| HGNC approved gene symbol | OMIM gene ID | % covered $\geq 10x$ | % covered $\geq 20x$ | % covered $\geq 30x$ | % covered $\geq 50x$ |
|---------------------------|--------------|----------------------|----------------------|----------------------|----------------------|
| WDR60                     | 615462       | 98.65                | 91.18                | 82.59                | 72.35                |
| WNT1                      | 164820       | 100                  | 100                  | 99.24                | 92.52                |
| WNT5A                     | 164975       | 100                  | 100                  | 97.90                | 94.92                |
| XRCC4                     | 194363       | 100                  | 100                  | 99.52                | 88.20                |
| XYLT1                     | 608124       | 98.64                | 94.96                | 91.15                | 83.65                |
| XYLT2                     | 608125       | 94.49                | 94.49                | 94.39                | 89.31                |
| ZBTB16                    | 176797       | 100                  | 100                  | 100                  | 100                  |
| ZMPSTE24                  | 606480       | 100                  | 100                  | 100                  | 97.48                |

- 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