Whole Exome Sequencing
Gene package Severe Combined Immune Deficiency (SCID)
Version 5, 25-2-2022

Technical information
DNA was enriched using the Agilent SureSelectXT Human All Exon V7 capture kit and paired-end sequenced on the Illumina platform (outsourced). Sequencing data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the hg19 genome and sequence variant detection. The detected sequence variants are annotated and filtered with Alissa Interpret software and classified with Alamut Visual. Copy number 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 annotated and filtered with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). The sensitivity to detect variants using this technology is not 100%; pathogenic variants could be missed. 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ADA	608958	100	100	99.94	92.54
AK2	103020	100	98.15	95.34	89.19
B2M	109700	100	100	100	100
CD247	186780	85.23	85.23	85.23	83.73
CD3D	186790	100	100	100	100
CD3E	186830	100	100	100	98.59
CD3G	186740	100	100	100	100
CD8A	186910	100	100	100	91.64
CIITA	600005	100	99.34	96.74	87.09
CORO1A	605000	92.29	91.79	90.59	86.61
DCLRE1C	605988	100	100	100	95.14
DOCK2	603122	100	99.82	99.04	97.48
DOCK8	611432	100	99.94	99.37	96.42
FCHO1	613437	100	97.57	91.61	80.80
FOXN1	600838	100	100	99.29	94.96
IL2RG	308380	100	100	100	97.91
IL7R	146661	100	100	100	100
JAK3	600173	100	99.87	98.64	94.01
LAT	602354	100	100	96.91	76.37
LCK	153390	100	99.43	96.43	86.11
LIG4	601837	96.16	96.16	96.16	96.16
NHEJ1	611290	100	100	100	98.53
PAX1	167411	100	98.03	83.02	72.50
PNP	164050	100	100	100	100

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PRKDC	600899	100	99.09	98.86	97.00
PTPRC	151460	100	99.32	96.86	90.29
RAC2	602049	100	100	100	98.85
RAG1	179615	100	100	100	100
RAG2	179616	100	100	100	100
RFX5	601863	100	100	100	99.01
RFXANK	603200	100	100	98.72	86.17
RFXAP	601861	100	100	98.29	95.32
RMRP	157660	No coverage data			
STK4	604965	100	100	98.04	95.21
TAP1	170260	100	100	98.17	90.89
TAP2	170261	100	98.11	94.82	88.28
TAPBP	601962	100	98.82	95.34	84.72
TTC7A	609332	100	100	99.67	92.91
ZAP70	176947	100	100	98.52	93.32

- OMIM release used: 23-9-2021
- The statistics above are based on a set of 104 samples
- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x