Whole Exome Sequencing
Gene package Severe Combined Immune Deficiency (SCID)
Version 3,1, 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. 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).

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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	99.88	92.24
AK2	103020	100	99.22	95.40	86.92
B2M	109700	100	100	100	100
CD247	186780	100	100	100	96.63
CD3D	186790	100	100	100	100
CD3E	186830	100	100	100	94.56
CD3G	186740	100	100	100	100
CD8A	186910	100	100	99.27	86.61
CIITA	600005	100	99.24	96.46	84.40
CORO1A	605000	92.29	91.91	90.49	85.72
DCLRE1C	605988	100	100	99.04	90.85
DOCK2	603122	100	99.54	98.91	95.43
DOCK8	611432	100	99.53	98.26	93.32
FCHO1	613437	100	98.00	92.67	77.84
FOXN1	600838	100	100	99.41	94.84
IL2RG	308380	100	100	99.61	90.29
IL7R	146661	100	100	100	100
JAK3	600173	100	99.67	98.07	91.75
LAT	602354	100	100	97.06	77.64
LCK	153390	100	99.58	95.98	82.06
LIG4	601837	96.16	96.16	96.16	96.16
NHEJ1	611290	100	100	100	96.58
PNP	164050	100	100	100	99.19
PRKDC	600899	99.75	98.97	98.40	94.68

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
PTPRC	151460	99.69	97.76	94.03	86.79
RAC2	602049	100	100	100	98.42
RAG1	179615	100	100	100	100
RAG2	179616	100	100	100	100
RFX5	601863	100	100	100	98.57
RFXANK	603200	100	100	98.24	85.32
RFXAP	601861	100	99.89	97.15	85.62
RMRP	157660	No coverage			
STK4	157660	100	97.06	96.04	88.85
TAP1	604965	100	99.79	97.84	86.65
TAP2	170260	100	98.55	95.30	87.73
TAPBP	170261	100	98.61	94.64	81.20
TTC7A	601962	100	100	99.05	90.00
ZAP70	609332	100	99.67	97.76	91.34

- 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 (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x