Whole Exome Sequencing
Gene package Severe Combined Immune Deficiency (SCID)
Version 2, 21-2-2020

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/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations 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)	median depth	% covered >10x	% covered >20x	% covered >30x
ADA	608958	85	100	100	96
AK2	103020	68	100	100	97
B2M	109700	94	100	100	100
CD247	186780	74	100	100	100
CD3D	186790	100	100	100	99
CD3E	186830	113	100	100	100
CD3G	186740	76	100	100	99
CD8A	186910	100	100	100	97
CIITA	600005	117	100	100	99
CORO1A	605000	122	92	92	91
DCLRE1C	605988	64	100	100	92
DOCK2	603122	82	100	100	97
DOCK8	611432	74	100	99	94
FOXN1	600838	136	100	100	100
IL2RG	308380	54	100	99	91
IL7R	146661	60	100	100	99
JAK3	600173	104	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
LAT	602354	91	100	100	97
LCK	153390	160	100	100	100
LIG4	601837	71	100	100	100
NHEJ1	611290	55	100	100	96
PNP	164050	70	100	100	97
PRKDC	600899	73	100	99	94
PTPRC	151460	73	100	99	95
RAC2	602049	99	100	100	100
RAG1	179615	101	100	100	100
RAG2	179616	71	100	100	100
RFX5	601863	100	100	100	98
RFXANK	603200	118	100	100	100
RFXAP	601861	148	100	100	100
RMRP	157660	No coverage data			
STK4	604965	82	100	100	99
TAP1	170260	125	100	100	100
TAP2	170261	106	100	100	100
TAPBP	601962	106	100	100	100
TTC7A	609332	110	100	100	99
ZAP70	176947	151	100	100	100

- OMIM release used: 8-9-2019
- The statistics above are based on a set of 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x