Whole Exome Sequencing
Gene package Severe Combined Immune Deficiency (SCID)
Version 3, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + 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 Alissa Interpret 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	161	100	100	100
AK2	103020	99	100	100	98
B2M	109700	131	100	100	100
CD247	186780	180	100	100	100
CD3D	186790	150	100	100	100
CD3E	186830	103	100	95	90
CD3G	186740	143	100	100	100
CD8A	186910	221	100	100	100
CIITA	600005	286	100	100	100
CORO1A	605000	287	92	92	92
DCLRE1C	605988	151	100	97	91
DOCK2	603122	118	100	100	100
DOCK8	611432	118	100	99	96
FCHO1	613437	208	100	100	100
FOXN1	600838	201	100	100	100
IL2RG	308380	88	100	100	97
IL7R	146661	105	100	100	98
JAK3	600173	266	100	100	100
LAT	602354	181	100	100	100
LCK	153390	183	100	100	100
LIG4	601837	86	100	97	93
NHEJ1	611290	108	100	100	96
PNP	164050	329	100	100	98
PRKDC	600899	99	97	94	89

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PTPRC	151460	62	93	84	76
RAC2	602049	233	100	100	100
RAG1	179615	199	100	100	100
RAG2	179616	130	100	100	100
RFX5	601863	180	100	100	100
RFXANK	603200	218	100	100	100
RFXAP	601861	136	100	100	100
RMRP	157660	No coverage data			
STK4	157660	97	100	95	81
TAP1	604965	172	100	100	100
TAP2	170260	166	100	100	100
TAPBP	170261	233	100	100	100
TTC7A	601962	192	100	100	100
ZAP70	609332	312	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