Whole Exome Sequencing
Gene package Severe Combined Immune Deficiency (SCID)
Version 1.1, 31-1-2020

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).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ADA	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700	608958	131	100	100	100
AK2	Reticular dysgenesis, 267500	103020	88	100	100	100
B2M	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600	109700	131	100	100	100
CD247	?Immunodeficiency 25, 610163	186780	122	100	100	100
CD3D	Immunodeficiency 19, 615617	186790	134	100	100	100
CD3E	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615	186830	145	100	100	100
CD3G	Immunodeficiency 17, CD3 gamma deficient, 615607	186740	121	100	100	100
CD8A	CD8 deficiency, familial, 608957	186910	149	100	100	100
CIITA	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300	600005	161	100	100	100
CORO1A	Immunodeficiency 8, 615401	605000	204	100	100	100
DCLRE1C	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450	605988	92	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
DOCK2	Immunodeficiency 40, 616433	603122	119	100	100	100
DOCK8	Hyper-IgE recurrent infection syndrome, 243700	611432	109	100	100	99
IL2RG	Combined immunodeficiency, moderate, 312863 Severe combined immunodeficiency, 300400	308380	110	100	100	100
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971	146661	94	100	100	100
JAK3	SCID, T-negative/B-positive type, 600802	600173	142	100	100	100
LAT	No OMIM phenotype	602354	136	100	100	100
LCK	?Immunodeficiency 22, 615758	153390	182	100	100	100
LIG4	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500	601837	114	100	100	100
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291	611290	91	100	100	100
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	122	100	100	100
PRKDC	Immunodeficiency 26, with or without neurologic abnormalities, 615966	600899	113	100	100	100
PTPRC	{Hepatitic C virus, susceptibility to}, 609532 Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971	151460	112	100	100	100
RAC2	Neutrophil immunodeficiency syndrome, 608203	602049	137	100	100	100
RAG1	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457	179615	150	100	100	100
RAG2	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457	179616	120	100	100	100
RFX5	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920	601863	142	100	100	100
RFXANK	MHC class II deficiency, complementation group B, 209920	603200	149	100	100	100
RFXAP	Bare lymphocyte syndrome, type II, complementation group D, 209920	601861	207	100	100	100
RMRP	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460	157660	No coverage data			
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868	604965	122	100	100	100
TAP1	Bare lymphocyte syndrome, type I, 604571	170260	169	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
TAP2	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis	170261	131	100	100	100
TAPBP	Bare lymphocyte syndrome, type I, 604571	601962	142	100	100	100
TTC7A	Gastrointestinal defects and immunodeficiency syndrome, 243150	609332	157	100	100	100
ZAP70	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840	176947	167	100	100	100

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 7 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 describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x
- % Covered 30x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 30x