Whole Exome Sequencing
Gene package Autoinflammatory diseases, version 4, 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. Copy 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 filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). Additionally, MPLA analysis was performed for PLCG2 (probemix P430, version A1; MRC Holland). 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
ACP5	171640	100	100	100	100
ADA2	607575	100	100	100	99.95
ADAM17	603639	100	98.56	95.42	88.36
ADAR	146920	98.45	98.45	98.45	97.45
ADGRE2	606100	100	99.83	97.94	93.64
AP1S3	615781	100	100	96.17	95.91
CARD14	607211	100	98.89	96.67	86.80
COPA	601924	100	100	99.61	96.71
DDX58	609631	100	100	98.96	95.47
DNASE1	125505	100	100	100	95.53
DNASE1L3	602244	100	100	100	96.68
DNASE2	126350	100	100	96.58	86
IFIH1	606951	100	100	100	97.67
IKZF1	603023	100	100	99.08	94.82
IL10	124092	100	100	100	99.13
IL10RA	146933	100	100	98.56	94.08

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
IL10RB	123889	100	94.66	93.72	89.42
IL1RN	147679	100	100	100	98.94
IL36RN	605507	100	100	97.61	83.67
LACC1	613409	100	100	100	96.31
LPIN2	605519	100	100	99.77	97.16
MEFV	608107	100	100	99.10	92.94
MVK	251170	100	100	100	96.47
NCSTN	605254	100	100	100	98.28
NLRC4	606831	100	100	100	99.81
NLRP1	606636	100	100	99.71	96.84
NLRP12	609648	100	100	99.26	95.07
NLRP3	606416	100	100	99.92	98.10
NOD2	605956	100	100	99.78	96.90
OTULIN	615712	91.89	85.62	85.62	80.64
PEPD	613230	100	100	98.29	89.21
PIK3CD	602839	100	99.94	99.44	97.51
PLCG2	600220	100	100	99.33	93.91
POMP	613386	99.08	85.08	82.78	80.85
PRKCD	176977	97.63	97.63	97.63	96.24
PSENEN	607632	100	100	98.62	89.26
PSMA3	176843	100	100	100	93.20
PSMB4	602177	100	100	98.66	92.97
PSMB8	177046	100	100	100	99.26
PSMB9	177045	100	100	98.84	89.70
PSMG2	609702	100	100	100	97.64
PSTPIP1	606347	100	100	99.89	88.43
RBCK1	610924	100	100	100	88.80
RIPK1	603453	100	100	98.60	92.41
RNASEH2A	606034	100	100	98.86	85.19
RNASEH2B	610326	100	91.35	91.35	91.35
RNASEH2C	610330	100	100	100	97.20
RNF31	612487	100	99.93	98.90	95.41
SAMHD1	606754	100	100	100	99.98
SLC29A3	612373	100	97.60	97.60	97.14
STING1	612374	100	100	100	88.46
TNFAIP3	191163	100	100	99.01	96.82
TNFRSF1A	191190	100	100	100	92.91
TREX1	606609	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
TRNT1	612907	99.83	90.53	89.36	83.15
UBA1	314370	100	99.76	98.07	90.22
WDR1	604734	100	99.83	98.30	97.17

- 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