Whole Exome Sequencing
Gene package Autoinflammatory diseases, version 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. Additionally, MPLA analysis was performed for PLCG2 (probemix P430, version A1; MRC Holland). 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
ADA2	607575	96	100	100	99
ADAM17	603639	67	100	99	94
ADGRE2	606100	144	100	99	95
AP1S3	615781	49	100	100	93
CARD14	607211	112	100	99	99
COPA	601924	75	100	100	99
DNASE2	126350	95	100	100	100
IL10	124092	120	100	100	99
IL10RA	146933	125	100	100	99
IL10RB	123889	69	100	100	97
IL1RN	147679	76	100	100	90
IL36RN	605507	82	100	100	100
LACC1	613409	62	100	98	93
LPIN2	605519	77	100	100	98
MEFV	608107	105	100	100	100
MVK	251170	100	100	100	100
NCSTN	605254	91	100	100	98
NLRC4	606831	83	100	100	97
NLRP1	606636	103	100	100	100
NLRP12	609648	135	100	100	100
NLRP3	606416	125	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
NOD2	605956	113	100	100	99
OTULIN	615712	70	97	91	82
PLCG2	600220	107	100	100	99
POMP	613386	45	100	100	87
PSENEN	607632	88	100	100	100
PSMA3	176843	53	100	99	87
PSMB4	602177	73	100	100	100
PSMB8	177046	124	100	100	100
PSMB9	177045	59	100	90	90
PSMG2	609702	58	100	99	86
PSTPIP1	606347	109	100	100	99
RBCK1	610924	135	100	100	99
RIPK1	603453	54	100	97	91
RNF31	612487	121	100	100	99
SLC29A3	612373	152	100	99	99
STING1	612374	122	100	100	100
TNFAIP3	191163	106	100	99	96
TNFRSF1A	191190	97	100	98	94
TRNT1	612907	68	100	99	92
WDR1	604734	77	100	99	94

- Gene symbols according HGCN
- 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