Whole Exome Sequencing
Gene package Autoinflammatory diseases, 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. 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
ACP5	171640	280	100	100	100
ADA2	607575	157	100	100	100
ADAM17	603639	98	99	94	88
ADAR	146920	163	100	100	100
ADGRE2	606100	220	100	100	100
AP1S3	615781	101	100	100	99
CARD14	607211	291	100	100	100
COPA	601924	119	100	100	99
DDX58	609631	74	97	90	83
DNASE1	125505	290	100	100	100
DNASE1L3	602244	108	100	100	99
DNASE2	126350	178	100	100	100
IFIH1	606951	70	99	91	80
IKZF1	603023	257	100	100	99
IL10	124092	156	100	100	100
IL10RA	146933	181	100	100	100
IL10RB	123889	86	100	98	96
IL1RN	147679	138	100	100	100
IL36RN	605507	192	100	100	100
LACC1	613409	72	100	98	91
LPIN2	605519	125	100	100	99
MEFV	608107	158	100	100	100
MVK	251170	207	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
NCSTN	605254	140	100	100	100
NLRC4	606831	127	100	97	94
NLRP1	606636	173	100	100	100
NLRP12	609648	246	100	100	100
NLRP3	606416	404	100	100	100
NOD2	605956	268	100	100	100
OTULIN	615712	81	98	91	78
PEPD	613230	216	100	100	100
PIK3CD	602839	284	100	100	100
PLCG2	600220	146	100	100	98
POMP	613386	76	85	85	85
PRKCD	176977	201	100	100	100
PSENEN	607632	150	100	100	100
PSMA3	176843	57	93	86	71
PSMB4	602177	141	100	100	100
PSMB8	177046	205	100	100	100
PSMB9	177045	206	100	100	100
PSMG2	609702	87	100	96	90
PSTPIP1	606347	257	100	100	100
RBCK1	610924	200	100	100	100
RIPK1	603453	112	100	96	95
RNASEH2A	606034	265	100	100	100
RNASEH2B	610326	60	100	99	89
RNASEH2C	610330	261	100	100	100
RNF31	612487	216	100	100	100
SAMHD1	606754	97	100	99	93
SLC29A3	612373	233	100	100	100
STING1	612374	214	100	100	100
TNFAIP3	191163	169	100	99	97
TNFRSF1A	191190	204	100	100	100
TREX1	606609	313	100	100	100
TRNT1	612907	47	90	86	74
WDR1	604734	239	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