Whole Exome Sequencing
Gene package Amyloidosis, version 1.2, 25-2-2022

Technical information
DNA was enriched using the Agilent SureSelectXT Human All Exon V7 capture kit and paired-end sequenced on the Illumina platform (outsourced). Sequencing data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the hg19 genome and sequence variant detection. The detected sequence variants are annotated and filtered with Alissa Interpret software and classified with Alamut Visual. Copy number 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 annotated and filtered with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). The sensitivity to detect variants using this technology is not 100%; pathogenic variants could be missed. 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
APOA1	107680	100	100	100	99.30
APOA2	107670	100	100	100	99.44
APOC2	608083	100	100	100	100
APOC3	107720	100	100	100	98.32
B2M	109700	100	100	100	100
CST3	604312	100	98.09	91.97	83.73
FGA	134820	100	100	100	97.42
GSN	137350	100	98.34	94.75	86.19
IL31RA	609510	100	100	100	98.57
LYZ	153450	100	100	100	100
OSMR	601743	100	100	100	99.92
TTR	176300	100	100	100	100

- OMIM release used: 18-2-2021
- The statistics above are based on a set of 104 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