Whole Exome Sequencing
Gene package Aneurysm, version 10, 30-9-2021

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). 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
AEBP1	189980	100	100	99.56	94.39
ABL1	102620	100	100	100	99.21
ACTA2	607513	100	100	100	99.30
ADAMTS19	602981	100	99.44	94.94	87.44
BGN	301870	100	100	100	94.65
COL1A1	120150	100	100	99.00	92.15
COL1A2	120160	100	99.09	96.75	86.41
COL3A1	120180	99.82	98.39	96.02	89.41
COL5A1	120215	100	99.17	97.94	93.25
COL5A2	120190	100	99.89	98.64	90.01
DCHS1	603057	100	100	100	99.83
EFEMP2	604633	100	99.97	97.06	84.43
ELN	130160	100	99.13	95.08	75.43
FBN1	134797	100	100	100	99.50
FBN2	612570	100	100	100	99.22
FLNA	300017	99.98	98.83	97.47	92.13
FOXE3	601094	83.06	73.13	61.67	40.89
GATA5	611496	100	99.39	96.00	78.11
HCN4	605206	100	99.32	97.77	88.70

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
HEY2	604674	100	100	100	98.47
IPO8	605600	100	100	99.54	96.46
LMOD1	602715	100	100	100	97.28
LOX	153455	100	100	100	100
LTBP3	602090	99.90	98.83	94.23	80.91
MAT2A	601468	100	100	100	100
MFAP5	601103	100	100	100	98.28
MYH11	160745	100	99.84	98.72	92.21
MYLK	600922	100	99.97	98.92	95.89
NOTCH1	190198	99.03	98.64	97.87	94.58
NPR3	108962	100	100	100	100
PLOD1	153454	100	99.54	96.86	87.82
PRKG1	176894	100	100	100	95.45
ROBO4	607528	100	100	99.88	96.30
SKI	164780	97.53	92.04	86.04	74.94
SLC2A10	606145	100	100	100	98.61
SMAD2	601366	100	100	100	98.69
SMAD3	603109	100	100	100	99.75
SMAD4	600993	100	100	100	99.36
SMAD6	602931	100	85.24	60.94	46.59
TGFB2	190220	100	100	100	95.93
TGFB3	190230	100	100	100	97.89
TGFBR1	190181	93.12	93.12	93.12	93.12
TGFBR2	190182	100	100	100	97.70
TLN1	186745	100	99.94	99.10	96.38

- OMIM release used: 23-9-2021
- The statistics above are based on a set of 150 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