Whole Exome Sequencing
Gene package Aneurysm, version 10.1, 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).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
AEBP1	189980	100	100	99.44	94.80
ABL1	102620	100	99.92	99.59	98.04
ACTA2	607513	100	100	100	98.06
ADAMTS19	602981	100	99.54	95.24	88.22
BGN	301870	100	100	100	96.78
COL1A1	120150	100	100	98.54	89.55
COL1A2	120160	100	99.02	95.78	84.61
COL3A1	120180	100	98.38	95.80	88.92
COL5A1	120215	100	99.39	98.11	92.79
COL5A2	120190	100	99.86	98.38	90.59
DCHS1	603057	100	100	100	99.67
EFEMP2	604633	100	99.25	95.45	79.59
ELN	130160	100	98.14	92.73	71.74
FBN1	134797	100	100	100	99.38
FBN2	612570	100	100	100	99.22
FLNA	300017	99.95	99.05	97.51	95.12
FOXE3	601094	82.50	73.08	63.20	37.82
GATA5	611496	100	99.62	96.45	79.18
HCN4	605206	100	99.75	97.81	88.26

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	99.19
IPO8	605600	100	100	99.99	96.65
LMOD1	602715	100	100	100	95.73
LOX	153455	100	100	100	100
LTBP3	602090	99.59	98.30	93.77	80.79
MAT2A	601468	100	100	100	100
MFAP5	601103	100	100	100	99.79
MYH11	160745	100	99.67	98.35	91.31
MYLK	600922	100	99.86	98.82	95.65
NOTCH1	190198	99.03	98.46	97.39	92.00
NPR3	108962	100	100	100	100
PLOD1	153454	100	99.25	95.92	85.89
PRKG1	176894	100	100	100	96.86
ROBO4	607528	100	100	99.44	93.12
SKI	164780	97.72	91.07	85.13	73.30
SLC2A10	606145	100	100	100	98.14
SMAD2	601366	100	100	100	98.09
SMAD3	603109	100	100	100	99.63
SMAD4	600993	100	100	100	99.47
SMAD6	602931	100	87.44	66.07	48.41
TGFB2	190220	100	100	100	95.05
TGFB3	190230	100	100	100	96.88
TGFBR1	190181	93.12	93.12	93.12	93.12
TGFBR2	190182	100	100	100	98.71
TLN1	186745	100	99.75	98.39	95.24

- OMIM release used: 23-9-2021
- 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