Whole Exome Sequencing
Gene package Aneurysm, version 8, 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. 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).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ABL1	189980	215	100	100	100
ACTA2	102620	142	100	100	100
ADAMTS19	607513	87	99	96	90
AEBP1	602981	276	100	100	100
BGN	301870	186	100	100	100
COL1A1	120150	246	100	100	100
COL1A2	120160	131	98	97	96
COL3A1	120180	113	100	98	95
COL5A1	120215	212	100	100	100
COL5A2	120190	108	99	99	98
DCHS1	603057	292	100	100	100
EFEMP2	604633	217	100	100	100
ELN	130160	171	100	100	100
FBN1	134797	141	100	100	99
FBN2	612570	139	100	100	99
FLNA	300017	206	100	100	100
FOXE3	601094	100	93	83	82
GATA5	611496	213	100	100	100
HCN4	605206	306	100	100	100
LMOD1	602715	134	100	100	100
LOX	153455	226	100	100	100
LTBP3	602090	223	100	99	99
MAT2A	601468	102	100	100	96
MFAP5	601103	96	100	96	85
MYH11	160745	171	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
MYLK	600922	195	100	100	100
NOTCH1	190198	318	100	99	99
NPR3	108962	260	100	100	98
PLOD1	153454	233	100	100	100
PRKG1	176894	84	98	91	89
ROBO4	607528	199	100	100	100
SKI	164780	208	100	100	98
SLC2A10	606145	216	100	100	100
SMAD2	601366	87	100	100	94
SMAD3	603109	239	100	100	100
SMAD4	600993	78	100	97	95
SMAD6	602931	188	100	100	100
TGFB2	190220	126	100	100	97
TGFB3	190230	182	100	100	100
TGFBR1	190181	110	97	94	92
TGFBR2	190182	214	100	100	99
TLN1	186745	195	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