Whole Exome Sequencing
Gene package Aneurysm, version 7, 21-2-2020

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/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ABL1	189980	141	100	100	100
ACTA2	102620	167	100	100	100
AEBP1	602981	142	100	100	100
BGN	301870	87	100	100	98
COL1A1	120150	130	100	100	100
COL1A2	120160	70	100	100	97
COL3A1	120180	122	100	100	100
COL5A1	120215	136	100	100	98
COL5A2	120190	65	100	100	94
DCHS1	603057	128	100	100	100
EFEMP2	604633	103	100	100	100
ELN	130160	85	100	100	98
FBN1	134797	186	100	100	100
FBN2	612570	74	100	100	97
FLNA	300017	110	100	100	100
FOXE3	601094	74	87	79	74

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
GATA5	611496	78	100	99	95
HCN4	605206	103	100	100	99
LMOD1	602715	128	100	100	100
LOX	153455	119	100	100	100
LTBP3	602090	130	100	99	96
MAT2A	601468	83	100	100	96
MFAP5	601103	50	100	100	92
MYH11	160745	136	100	100	99
MYLK	600922	121	100	100	99
NOTCH1	190198	117	100	99	98
PLOD1	153454	100	100	100	99
PRKG1	176894	69	100	100	98
ROBO4	607528	91	100	100	98
SKI	164780	123	100	100	99
SLC2A10	606145	125	100	100	100
SMAD2	601366	69	100	100	97
SMAD3	603109	188	100	100	100
SMAD4	600993	79	100	100	98
SMAD6	602931	165	100	97	87
TGFB2	190220	88	100	100	98
TGFB3	190230	104	100	100	100
TGFBR1	190181	171	94	93	93
TGFBR2	190182	210	100	100	100
TLN1	186745	100	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