Whole Exome Sequencing
Gene package Pulmonary Arterial Hypertension (PAH)
version 1.2, 26-2-2021

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
ABCC8	600509	100	100	100	97.41
ACVRL1	601284	100	100	98.82	85.20
AQP1	107776	100	100	98.65	91.88
ATP13A3	610232	100	98.53	96.99	89.43
BMP10	608748	100	100	100	100
BMPR1B	603248	100	100	97.99	91.96
BMPR2	600799	100	100	99.44	96.27
CAV1	601047	100	100	97.64	91.58
CBLN2	600433	100	95.83	92.35	84.43
CPS1	608307	99.28	99.28	99.28	98.64
EIF2AK4	609280	100	99.46	97.95	92.74
ENG	131195	100	100	96.77	87.73
GDF2	605120	100	100	99.47	96.91
GGCX	137167	100	100	100	89.80
KCNA5	176267	100	99.46	95.48	84.37
KCNK3	603220	100	96.11	92.92	81.51
KLF2	602016	92.44	83.69	71.64	45.87
KLK1	147910	100	100	100	97.52
MMACHC	609831	100	100	100	95.84
NOTCH3	600276	99.47	95.64	92.41	82.85
SARS2	612804	100	100	99.50	91.61
SMAD1	601595	100	100	99.80	94.29
SMAD4	600993	100	100	100	98.00

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SMAD5	603110	100	100	100	98.37
SMAD9	603295	100	99.31	96.15	90.73
SOX17	610928	100	100	99.80	92.90
TBX4	601719	100	96.41	92.80	82.15
TOPBP1	607760	99.97	99.53	98.64	94.80

- OMIM release used: 18-2-2021
- The statistics above are based on a set of 100 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