Whole Exome Sequencing
Gene package Pulmonary Arterial Hypertension (PAH)
version 1.4, 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).

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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	98.90
ACVRL1	601284	100	100	100	88.28
AQP1	107776	100	100	98.48	92.33
ATP13A3	610232	100	99.82	98.41	94.07
BMP10	608748	100	100	100	100
BMPR1B	603248	100	100	100	96.53
BMPR2	600799	100	100	100	97.15
CAV1	601047	100	100	98.32	91.67
CBLN2	600433	100	96.58	93.51	87.09
CPS1	608307	99.28	99.28	99.28	99.15
EIF2AK4	609280	100	100	99.35	96.17
ENG	131195	100	100	98.31	90.90
GDF2	605120	100	100	99.77	96.99
GGCX	137167	100	100	100	93.20
KCNA5	176267	100	99.76	96.29	86.82
KCNK3	603220	100	97.05	94.27	86.21
KLF2	602016	91.11	82.53	71.33	44.71
KLK1	147910	100	100	100	98.42
MMACHC	609831	100	100	100	95.30
NOTCH3	600276	99.58	95.67	92.96	85.47
SARS2	612804	100	100	99.95	95.16
SMAD1	601595	100	100	99.97	96.14
SMAD4	600993	100	100	100	99.47

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SMAD5	603110	100	100	100	100
SMAD9	603295	100	99.38	96.78	91.72
SOX17	610928	100	100	100	97.39
TBX4	601719	100	98.25	94.05	87.49
TOPBP1	607760	100	99.76	99.35	96.66

- OMIM release used: 18-2-2021
- 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