Whole Exome Sequencing
Gene package Pulmonary Arterial Hypertension (PAH)
version 1.1, 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. Duplicateand 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ABCC8	600509	242	100	100	100
ACVRL1	601284	298	100	100	100
AQP1	107776	216	100	100	100
ATP13A3	610232	56	98	91	78
BMP10	608748	178	100	100	100
BMPR1B	603248	79	100	98	92
BMPR2	600799	102	100	100	98
CAV1	601047	134	100	100	100
CBLN2	600433	159	100	100	99
CPS1	608307	101	100	100	99
EIF2AK4	609280	110	98	95	91
ENG	131195	281	100	100	100
GDF2	605120	204	100	100	100
GGCX	137167	113	100	100	97
KCNA5	176267	336	100	100	100
KCNK3	603220	231	100	100	98
KLF2	602016	149	100	95	90
KLK1	147910	238	100	100	100
MMACHC	609831	205	100	100	100
NOTCH3	600276	291	100	100	99
SARS2	612804	272	100	100	100
SMAD1	601595	129	100	100	100
SMAD4	600993	78	100	97	95
SMAD5	603110	100	100	97	94
SMAD9	603295	117	100	99	97
SOX17	610928	198	100	100	100

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TBX4	601719	204	100	100	100
TOPBP1	607760	85	97	92	85

- 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