

Whole Exome Sequencing

Gene package Idopathic Pulmonary Fibrosis, version 6, 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).



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ABCA3	601615	99.96	99.00	97.58	89.59
AP3B1	603401	97.72	95.91	94.30	84.26
COPA	601924	100	100	99.61	96.71
DKC1	300126	100	98.05	94.16	83.85
FAM111B	615584	100	100	100	99.55
GFRA1	601496	100	100	97.81	90.85
MARS1	156560	100	100	99.29	95.58
MUC5B	600770	99.95	99.17	97.16	89.13
NKX2-1	600635	100	100	97.55	73.71
NOP10	606471	100	100	100	100
OAS1	164350	100	97.94	95.89	87.63
PARN	604212	100	100	100	94.26
RTEL1	608833	100	100	99.24	90.23
SERPINA1	107400	100	100	100	100
SFTPA2	178642	100	100	100	99.77
SFTPC	178620	100	100	98.19	92.33
STING1	612374	100	100	100	88.46
TERC	602322	No coverage data			
TERT	187270	100	98.75	95.08	87.33
TINF2	604319	100	100	100	99.35
ZCCHC8	616381	100	98.42	96.73	91.48

- 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