Whole Exome Sequencing
Gene package Idopathic Pulmonary Fibrosis, version 8, 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
ABCA3	601615	99.81	99.01	97.90	91.35
ADAR	146920	98.45	98.45	98.45	98.04
AP3B1	603401	99.05	96.39	94.98	89.23
AP3D1	607246	97.71	97.64	95.96	89.40
ASAH1	613468	100	100	99.41	95.80
BLOC1S3	609762	100	100	91.37	52.16
BLOC1S5	607289	100	100	98.18	77.46
BLOC1S6	604310	100	100	96.91	78.29
CCDC103	614677	100	100	100	100
CCDC39	613798	100	99.71	98.09	87.36
CCDC40	613799	100	99.45	96.22	89.22
CCDC65	611088	100	100	99.69	93.92
CCNO	607752	100	100	100	95.50
CDKN2A	600160	100	100	96.17	86.99
CFAP221	618704	100	100	99.97	97.42
CFAP298	615494	100	100	100	98.92
CFAP300	618058	100	100	100	89.80
CFAP52	609804	100	100	100	99.11
COPA	601924	100	100	100	98.67
CSF2RA	306250	100	100	100	100
CSF2RB	138981	100	98.32	94.93	88.92

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CTC1	613129	100	100	99.71	94.64
DKC1	300126	100	100	98.05	93.43
DNAAF1	613190	100	100	100	96.17
DNAAF2	612517	100	100	100	100
DNAAF3	614566	100	99.98	97.12	86.12
DNAAF4	608706	100	96.77	92.99	82.50
DNAAF5	614864	94.87	84.62	79.17	73.54
DNAAF6	300933	100	100	100	99.54
DNAH11	603339	100	100	99.94	98.42
DNAH5	603335	100	99.85	99.34	97.23
DNAH8	603337	100	99.96	99.48	96.00
DNAH9	603330	100	100	99.62	97.55
DNAI1	604366	100	100	99.28	94.36
DNAI2	605483	93.11	93.11	92.91	91.19
DNAJB13	610263	100	100	100	100
DNAL1	610062	100	100	89.73	89.59
DRC1	615288	100	100	99.82	95.25
DTNBP1	607145	100	100	100	96.72
ERF	611888	100	100	98.23	86.34
FAM111B	615584	100	100	100	100
FARSA	602918	100	100	99.83	95.35
FARSB	609690	100	100	100	98.74
FGF10	602115	100	100	100	96.35
FGFR2	176943	100	100	99.68	98.15
FLNA	300017	99.95	99.05	97.51	95.12
FOXF1	601089	100	100	100	93.67
FOXJ1	602291	100	100	97.16	90.48
GAS2L2	611398	100	100	100	97.57
GAS8	605178	100	100	100	94.33
GATA2	137295	100	100	100	85.78
GBA	606463	100	100	100	99.29
GFRA1	601496	100	100	99.59	94.48
HPS1	604982	100	100	99.68	90.74
HPS3	606118	100	100	98.87	91.36
HPS4	606682	100	100	100	96.66
HPS5	607521	100	100	99.35	95.43
HPS6	607522	100	100	98.59	84.20
HYDIN	610812	98.12	90.57	83.20	69.56
INVS	243305	100	99.60	98.89	96.61
ITGA3	605025	100	100	98.99	94.16
LRRC56	618227	100	100	99.35	82.50
LRRC57	No OMIM ID	100	100	100	100

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MARS1	156560	100	100	99.63	97.32
MCIDAS	614086	100	100	100	98.26
MUC5B	600770	99.90	99.06	96.87	89.33
NEK10	618726	100	100	99.39	96.91
NHP2	606470	100	100	100	100
NKX2-1	600635	100	100	99.96	81.75
NME7	613465	100	100	99.93	94.23
NME8	607421	100	100	98.69	89.41
NOP10	606471	100	100	100	100
NPC1	607623	100	99.98	98.96	97.40
NPC2	601015	100	100	100	100
NSMCE3	608243	100	100	100	95.60
OAS1	164350	100	97.91	95.56	87.22
ODAD1	615038	96.25	96.08	93.98	84.17
ODAD2	615408	100	98.60	96.70	92.34
ODAD3	615956	100	99.34	97.48	90.24
ODAD4	617095	100	100	99.75	91.36
PARN	604212	100	100	100	98.33
PEPD	613230	100	100	98.93	91.34
PLCG2	600220	100	100	100	96.79
PSAP	176801	100	100	100	95.51
RASGRP1	603962	100	100	100	99.95
RNASE2	131410	100	100	100	100
RNASEH2B	610326	100	91.35	91.35	91.35
RNF168	612688	100	100	100	99.65
RSPH1	609314	100	100	99.08	87.36
RSPH3	615876	100	100	100	96.83
RSPH4A	612647	100	100	100	100
RSPH6A	607548	100	97.56	92.10	80.54
RTEL1	608833	100	100	99.77	94.04
SAMHD1	606754	100	100	100	100
SERPINA1	107400	100	100	100	100
SFTPA1	178630	100	100	100	99.83
SFTPA2	178642	100	100	100	100
SFTPB	178640	100	100	100	98.59
SFTPC	178620	100	100	98.41	92.98
SLC34A2	604217	100	100	100	98.12
SLC7A7	603593	100	100	100	99.01
SMPD1	607608	100	100	100	98.58
SPAG1	603395	100	98.04	93.83	80.54
SPEF2	610172	100	100	98.57	95.14
STAT3	102582	100	100	100	95.99

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STING1	612374	100	100	100	88.83
STK36	607652	100	99.94	99.43	98.30
STRA6	610745	100	100	100	96.01
TBX4	601719	100	98.25	94.05	87.49
TERC	602322	No coverage data			
TERT	187270	100	99.15	96.14	90.14
TINF2	604319	100	100	100	100
TREX1	606609	100	100	100	100
TTC12	610732	100	100	100	97.88
UBA1	314370	100	99.93	97.88	90.57
USB1	613276	100	99.20	88.66	75.29
ZCCHC8	616381	100	99.19	97.81	95.27
ZMYND10	607070	100	100	100	100

- OMIM release used: 23-9-2021
- 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