

# Whole Exome Sequencing

## Gene package Idopathic Pulmonary Fibrosis, version 5.1, 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. 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/>). 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).



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ABCA3	601615	281	100	100	100
AP3B1	603401	58	93	79	66
COPA	601924	119	100	100	99
DKC1	300126	73	100	93	88
FAM111B	615584	98	100	98	94
GFRA1	601496	183	100	100	99
MARS1	156560	161	100	100	100
MUC5B	600770	279	100	100	100
NKX2-1	600635	216	100	100	100
OAS1	164350	131	100	100	100
PARN	604212	79	100	98	93
RTEL1	608833	260	100	100	100
SERPINA1	107400	176	100	100	100
SFTPA2	178642	255	100	100	100
SFTPC	178620	186	100	100	100
STING1	612374	214	100	100	100
TERC	602322	No coverage data			
TERT	187270	276	100	100	99
TINF2	604319	229	100	100	100

- 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 ( $\pm 10$ bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence ( $\pm 10$ bp flanking introns) that is covered at least 10x, 20x or 30x