

Whole Exome Sequencing

Gene package Idiopathic Pulmonary Fibrosis, version 5, 21-2-2020



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/>). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: <http://www.broadinstitute.org/gatk/>). The detected variants are filtered and annotated with Cartagenia 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	107	100	100	98
AP3B1	603401	66	100	98	87
COPA	601924	75	100	100	99
DKC1	300126	53	100	98	89
FAM111B	615584	65	100	100	99
GFRA1	601496	86	100	100	97
MARS1	156560	101	100	100	100
MUC5B	600770	184	100	100	99
NKX2-1	600635	95	100	100	100
OAS1	164350	108	100	100	100
PARN	604212	59	100	99	92
RTEL1	608833	131	100	100	99
SERPINA1	107400	90	100	100	100
SFTP2	178642	191	100	100	100
SFTPC	178620	100	100	100	100
STING1	612374	122	100	100	100
TERC	602322	No coverage data			
TERT	187270	136	100	100	99
TINF2	604319	155	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 (± 10 bp flanking introns) of the longest transcript
- % Covered 10x, 20x and 30x describes the percentage of a gene's coding sequence (± 10 bp flanking introns) that is covered at least 10x, 20x or 30x