Whole Exome Sequencing
Gene package Idopathic Pulmonary Fibrosis, version 4, 18-2-2019

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate 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).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	Transcript	median depth	% covered >10x	% covered >20x	% covered >30x
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3, 610921	601615	NM_001089.2	104	100	100	97
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	NM_003664.4	57	100	96	80
COPA	{Autoimmune interstitial lung, joint, and kidney disease}, 616414	601924	NM_001098398.1	62	100	100	96
DKC1	Dyskeratosis congenita, X-linked, 305000	300126	NM_001363.4	43	100	97	79
FAM111B	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704	615584	NM_198947.3	56	100	100	96
GFRA1	No OMIM phenotype	601496	NM_005264.5	79	100	99	94
MARS	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486	156560	NM_004990.3	90	100	100	100
MUC5B	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500	600770	NM_002458.2	173	100	100	99
NKX2-1	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550	600635	NM_003317.3	89	100	100	100
PARN	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371	604212	NM_002582.3	52	100	99	88
RTEL1	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	NM_001283009.1	132	100	100	0
SERPINA1	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963	107400	NM_000295.4	79	100	100	99
SFTPA2	Pulmonary fibrosis, idiopathic, 178500	178642	NM_001098668.3	209	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	Transcript	median depth	% covered >10x	% covered >20x	% covered >30x
SFTPC	Surfactant metabolism dysfunction, pulmonary, 2, 610913	178620	NM_003018.3	94	100	100	100
TERC	{Aplastic anemia}, 614743 Dyskeratosis congenita, autosomal dominant 1, 127550 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743	602322	NR_001566.1	No coverage data			
TERT	{Dyskeratosis congenita, autosomal dominant 2}, 613989 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742	187270	NM_198253.2	131	100	100	99
TINF2	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130	604319	NM_001099274.1	140	100	100	100
TMEM173	STING-associated vasculopathy, infantile-onset, 615934	612374	NM_198282.3	116	100	100	100

- Gene symbols according HGCN
- OMIM release used: 4-7-2018
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 95 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 describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x
- % Covered 30x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 30x