Whole Exome Sequencing
Gene package Idopathic Pulmonary Fibrosis, version 1, 1-7-2017

Technical information
After DNA was enriched using Agilent Sureselect Clinical Research Exome (CRE) Capture, samples were run on the Illumina Hiseq platform. The aim is to obtain 50 million total reads per exome with a mapped fraction >0.98. The average coverage of the exome is ~50x. Data are demultiplexed by Illumina software bcl2fastq. Reads are mapped to the genome using BWA (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by Genome Analysis Toolkit (reference: http://www.broadinstitute.org/gatk/). Analysis is performed in Cartagenia using The Variant Calling File (VCF) followed by filtering. 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).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	Transcript	median depth	% covered >10x	% covered >20x
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3, 610921	601615	NM_001089.2	87	100	100
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	NM_003664.4	73	100	95
COPA	{Autoimmune interstitial lung, joint, and kidney disease}, 616414	601924	NM_001098398.1	42	98	86
DKC1	Dyskeratosis congenita, 305000	300126	NM_001363.4	57	100	94
FAM111B	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704	615584	NM_198947.3	61	99	94
GFRA1	No OMIM phenotype	601496	NM_005264.4	74	100	99
MUC5B	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500	600770	NM_002458.2	173	100	99
NKX2-1	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550	600635	NM_003317.3	70	100	97
PARN	Dyskeratosis congenita 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371	604212	NM_002582.3	45	97	78
SFTPA2	Pulmonary fibrosis, idiopathic, 178500	178642	NM_001098668.2	68	100	100
SFTPC	Surfactant metabolism dysfunction, pulmonary, 2, 610913	178620	NM_003018.3	84	100	100
TERC	{Aplastic anemia}, 614743 Dyskeratosis congenita 1, 127550 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743	602322	NR_001566.1	no coverage data		

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	Transcript	median depth	% covered >10x	% covered >20x
TERT	{Dyskeratosis congenita 2}, 613989 {Dyskeratosis congenita 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	83	96	94
TINF2	Dyskeratosis congenita 3, 613990 Revesz syndrome, 268130	604319	NM_001099274.1	92	100	100
TMEM173	STING-associated vasculopathy, infantile-onset, 615934	612374	NM_198282.2	77	100	100

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "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 50 samples
- Median depth is the median of the mean sequence depth over the protein coding exons of the transcript
- % Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x