Supplement 1.

Gene	Associated phenotypes	Inheritan	ClinVar	HGMD
ABCA3	Interstitial lung disease, Surfactant metabolism dysfunction, pulmonary	AD/AR	11	287
ARHGEF1	Idiopathic bronchiectasis, Immunodeficiencies with antibody defects	AR		1
C11ORF70	Primary ciliary dyskinesia	AR		5
CCDC39	Ciliary dyskinesia	AR	39	47
CCDC40	Ciliary dyskinesia	AR	33	43
CFTR	Cystic fibrosis, Congenital bilateral absence of the vas deferens	AD/AR	518	1803
CHAT	Myasthenic syndrome, congenital	AR	24	73
CHRNA1	Myasthenic syndrome, congenital	AD/AR	28	35
CHRNB1	Myasthenic syndrome	AD/AR	11	11
CHRND	Myasthenic syndrome	AD/AR	18	26
CHRNE	Myasthenic syndrome	AD/AR	48	134
COLQ	Myasthenic syndrome, congenital	AR	23	67
CSF2RA#*	Surfactant metabolism dysfunction, pulmonary	XL	2	17
CSF2RB	Surfactant metabolism dysfunction, pulmonary, 5	AR	2	6
DKC1	Hoyeraal-Hreidarsson syndrome, Dyskeratosis congenita	XL	48	74
DNAAF1	Ciliary dyskinesia	AR	19	38
DNAAF2	Ciliary dyskinesia	AR	13	6
DNAH1	Spermatogenic failure 18	AR	15	32
DNAH11*	Ciliary dyskinesia	AR	66	130
DNAH5	Ciliary dyskinesia	AR	140	197
DNAH9	Primary ciliary dyskinesia	AR		6
DNAI1	Ciliary dyskinesia	AR	17	35
DNAI2	Ciliary dyskinesia	AR	19	6
DNAL1	Ciliary dyskinesia	AR	3	1
EDN3	Hirschsprung disease, Central hypoventilation syndrome, congenital, Waardenburg syndrome	AD/AR	7	21
EFEMP2	Cutis laxa	AR	14	16
ELMOD2	Familial idiopathic pulmonary fibrosis	AD/AR		
ELN	Cutis laxa, Supravalvular aortic stenosis	AD	78	113
FAM111B*	Hereditary Fibrosing Poikiloderma with Tendon Contracture, Myopathy, and Pulmonary Fibrosis, Lung cancer, familial, susceptibilty to	AD	7	7
FBLN5	Cutis laxa, Macular degeneration, age-related	AD/AR	13	22
FLCN	Birt-Hogg-Dube syndrome, Pneumothorax, primary spontaneous	AD	154	210
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins	AD	10	102
GAS2L2	Primary ciliary dyskinesia	AR		3
GAS8	Ciliary dyskinesia, primary, 33	AR	4	6
GLRA1	Hyperekplexia	AD/AR	39	69
HPS1*	Hermansky-Pudlak syndrome	AR	28	55
HPS4	Hermansky-Pudlak syndrome	AR	16	22
ITGA3	Interstitial lung disease with nephrotic syndrome and epidermolysis bullosa	AR	6	11
LTBP4	Cutis laxa with severe pulmonary, gastrointestinal, and urinary abnormalities	AR	10	17
MCIDAS	Primary ciliary dyskinesia	AR	4	3
MECP2	Angelman-like syndrome, Autism, Rett syndrome, Encephalopathy, Mental retardation	XL	506	1039
NAF1		AD		2
NF1*	Watson syndrome, Neurofibromatosis, Neurofibromatosis-Noonan syndrome	AD	1157	2901
NKX2-1	Thyroid cancer, nonmedullary, Choreoathetosis, hypothyroidism, and neonatal respiratory distress, Chorea, hereditary benign	AD	27	137
NME8	Ciliary dyskinesia	AR	1	6
PARN*	Pulmonary fibrosis and/or bone marrow failure, Dyskeratosis congenita	AD/AR	15	29
PHOX2B	Central hypoventilation syndrome, congenital, Neuroblastoma, susceptiblity to, Neuroblastoma with Hirschsprung disease	AD	11	86
PIH1D3	Ciliary dyskinesia, primary, 36	XL	2	12
POLD1	Colorectal cancer, Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, Idiopathic bronchiectasis, Immunodeficiency	AD/AR	3	31
RAPSN	Myasthenic syndrome, congenital	AR	26	58
RET	Hirschsprung disease, Central hypoventilation syndrome, congenital, Pheochromocytoma, Medullary thyroid carcinoma, Multiple endocrine neoplasia	AD/AR	122	407
RSPH3	Ciliary dyskinesia, primary, 32	AR	7	5
RSPH4A	Ciliary dyskinesia	AR	18	24
RSPH9	Ciliary dyskinesia	AR	8	12
RTEL1	Pulmonary fibrosis and/or bone marrow failure, Dyskeratosis congenita	AD/AR	58	51
SCN4A	Hyperkalemic periodic paralysis, Myotonia, potassium-aggravated, Paramyotonia congenita, Myasthenic syndrome, congenital, Normokalemic potassium-sensitive periodic paralysis	AD/AR	57	126
SCNN1A	Pseudohypoaldosteronism, Bronchiectasis with or without elevated sweat chloride	AD/AR	10	44
SCNN1B	Liddle syndrome, Pseudohypoaldosteronism, Bronchiectasis with or without elevated sweat chloride	AD/AR	19	47
SERPINA1	Alpha-1-antitrypsin deficiency	AR	49	80
SFTPA1	Idiopathic pulmonary fibrosis	AD		2
SFTPA2	Pulmonary fibrosis, idiopathic	AD	2	5
SFTPB	Surfactant metabolism dysfunction, pulmonary	AR	5	28
SFTPC	Surfactant metabolism dysfunction, pulmonary	AD	8	82
SLC34A2	Pulmonary alveolar microlithiasis	AR	5	19
SLC6A5	Hyperekplexia	AR	15	33
SLC7A7	Lysinuric protein intolerance	AR	55	67
SMPD1	Niemann-Pick disease	AR	110	249
STAT3	Hyper-IgE recurrent infection syndrome, Autoimmune disease, multisystem, infantile onset	AD	47	152
STK36	Primary ciliary dyskinesia	AR		5
STRA6	Microphthalmia, syndromic, Microphthalmia, isolated, with coloboma	AR	22	33
TERC	Aplastic anemia, Pulmonary fibrosis and/or bone marrow failure, telomere-related, Dyskeratosis congenita	AD	42	73
TERT	Aplastic anemia, Pulmonary fibrosis and/or bone marrow failure, telomere-related, Dyskeratosis congenita	AD/AR	48	156
TINF2	Revesz syndrome, Dyskeratosis congenita	AD	25	42
TMEM173	STING-associated vasculopathy, infantile-onsent (SAVI)	AD	4	10
TSC1	Lymphangioleiomyomatosis, Tuberous sclerosis	AD	177	372
TSC2	Lymphangioleiomyomatosis, Tuberous sclerosis	AD	396	1195
ZEB2*	Mowat-Wilson syndrome	AD	154	287
* Some, or all, of the gene is duplicated in the genome. Read more.				

The gene has suboptimal coverage (means <90% of the gene's target nucleotides are covered at >20x with mapping quality score (MQ>20) reads), and/or the gene has exons listed under Test limitations section that are not included in the panel

The sensitivity to detect variants may be limited in genes marked with an asterisk (*) or number sign (#)

Gene refers to the HGNC approved gene symbol; Inheritance refers to inheritance patterns such as autosomal dominant (AD), autosomal recessive (AR), mitochondrial (mi), X-linked (XL), X-linked dominant (XLD) and X-linked recessive (XLR); ClinVar refers to the number of variants in the gene classified as pathogenic or likely pathogenic in this database (ClinVar); HGMD refers to the number of variants with possible disease association in the gene listed in Human Gene Mutation Database (HGMD). The list of associated, gene specific phenotypes are generated from CGD or Mitomap databases.