PulmoGene Panel (64 Genes)				
Gene	Inheritance Pattern	Association		Syndromes/Clinical Features
		Non-Synd	Synd	
ABCA3	AR	X		Surfactant dysfunction, Pulmonary fibrosis
ACVRL1	AD	X		Pulmonary hypertension, Hereditary hemorrhagic telangiectasia
AP3B1	AR		X	Hermansky-Pudlak syndrome
ASCL1	AD		X	Central hypoventilation syndrome
BDNF	AD		X	Central hypoventilation syndrome
BLOC1S3	AR		X	Hermansky-Pudlak syndrome
BLOC1S6	AR		X	Hermansky-Pudlak syndrome
BMPR2	AD	X		Pulmonary arterial hypertension
CCDC39	AR		X	Primary ciliary dyskinesia, Bronchiectasis
CCDC40*	AR		X	Primary ciliary dyskinesia, Bronchiectasis
CFTR	AR	X		Cystic fibrosis, Bronchiectasis
CSF2RA*	XL	X		Familial pulmonary alveolar proteinosis
CSF2RB	Unkn	X		Familial pulmonary alveolar proteinosis
DNAAF1	AR		X	Primary ciliary dyskinesia, Bronchiectasis
DNAAF2	AR		X	Primary ciliary dyskinesia, Bronchiectasis
DNAH11	AR		X	Primary ciliary dyskinesia, Bronchiectasis
DNAH5	AR		X	Primary ciliary dyskinesia, Bronchiectasis
DNAI1	AR		X	Primary ciliary dyskinesia, Bronchiectasis
DNAI2	AR		X	Primary ciliary dyskinesia, Bronchiectasis
DNAL1	AR		X	Primary ciliary dyskinesia, Bronchiectasis
DOCK8	AR		X	Atopic & eosinophilic disease, Hyper-IgE recurrent infection syndrome
DTNBP1	AR		X	Hermansky-Pudlak syndrome
EDN3	AD		X	Central hypoventilation syndrome
EFEMP2	AR		X	Cutis laxa & emphysema
ELMOD2	Unkn	X		Pulmonary fibrosis
ELN	AD		X	Cutis laxa & emphysema
ENG	AD	X		Pulmonary hypertension, Hereditary hemorrhagic telangiectasia
FBLN5	AR / AD		X	Cutis laxa & emphysema
FBN1	AD		X	Neonatal Marfan (pulmonary emphysema), Marfan syndrome (pneumothorax)
FLCN	AD		X	Birt-Hogg-Dubé syndrome, Pneumothorax
FOXF1	AD	X		Alveolar capillary dysplasia
GDNF	AD		X	Central hypoventilation syndrome
HPS1	AR		X	Hermansky-Pudlak syndrome
HPS3	AR		X	Hermansky-Pudlak syndrome
HPS4	AR		X	Hermansky-Pudlak syndrome
HPS5	AR		X	Hermansky-Pudlak syndrome
HPS6	AR		X	Hermansky-Pudlak syndrome
HRAS	AD		X	Costello syndrome
LTBP4	AR		X	Cutis laxa & emphysema
MUC5B*	Risk	X		Pulmonary fibrosis (rs35705950- risk allele)
NF1	AD		X	Neurofibromatosis 1
NKX2-1	AD		X	Neonatal Respiratory Distress (Brain-Lung-Thyroid)
NME8	AR		X	Primary ciliary dyskinesia, Bronchiectasis
PHOX2B	AD		X	Central hypoventilation syndrome
RET	AD		X	Central hypoventilation syndrome
RPGR*	XL	X		Recurrent pulmonary infections
RSPH1	AR		X	Primary ciliary dyskinesia, Bronchiectasis
RSPH4A	AR		X	Primary ciliary dyskinesia, Bronchiectasis
RSPH9	AR		X	Primary ciliary dyskinesia, Bronchiectasis
SCNN1A	AR	X		Pseudohypoaldosteronism type 1, Bronchiectasis
SCNN1B	AR	X		Pseudohypoaldosteronism type 1, Bronchiectasis
SCNN1G	AR	X		Pseudohypoaldosteronism type 1, Bronchiectasis
SERPINA1	AR		X	Alpha-1 antitrypsin deficiency disorder
SFTPA1	Unkn	X		Pulmonary fibrosis
SFTPA2	Unkn	X		Pulmonary fibrosis
SFTPB	AD, AR	X		Surfactant dysfunction, Pulmonary fibrosis
SFTPC	AD	X		Surfactant dysfunction, Pulmonary fibrosis
SFTPD	Unkn	X		Surfactant dysfunction, Pulmonary fibrosis
SMAD9	AD	X		Pulmonary arterial hypertension
STAT3	AD		X	Autosomal dominant hyper-IgE syndrome
TERC	AD	X		Telomere shortening syndromes (Pulmonary fibrosis, Dyskeratosis congenita)
TERT	AD	X		Telomere shortening syndromes (Pulmonary fibrosis, Dyskeratosis congenita)
TSC1	AD		X	Tuberous sclerosis, Lymphangioleiomyomatosis
TSC2	AD		X	Tuberous sclerosis, Lymphangioleiomyomatosis

*Please note a few exons

** Inher. = Inheritance