Bronchiectasis and Primary Ciliary Dyskinesia Precision Panel

Overview

Bronchiectasis is a chronic lung disease characterized by a pathologic and irreversible dilation of the airways. The heterogeneity of bronchiectasis is a major challenge in clinical practice. There are numerous underlying causes of bronchiectasis, although in many cases no cause is found. Known causes include post-infectious, aspiration syndromes, defects in host defence, cystic fibrosis, primary ciliary dyskinesia or even be systemic such as common variable immunodeficiency and anatomical defects including intraluminal airway obstruction, intramural obstruction or external airway compression. Bronchiectasis can be seen in all age groups, but the highest prevalence of disease is seen in the older age range (greater than 60) and women are disproportionately affected.

Primary Ciliary Dyskinesia (PCD) is a genetically and clinically heterogeneous disorder of motile cilia causing failure of mucociliary clearance and organ laterality defects and infertility inherited in an autosomal recessive pattern. It belongs to a rapidly expanding collection of disorders known as ciliopathies. Patients with primary ciliary dyskinesia have diverse clinical manifestations, including chronic upper and lower respiratory tract disease, left-right laterality defects, and infertility. A growing number of disease-associated genes and pathogenic mutations have been identified which encode ciliary structures that allow cilia to be functionally motile.

The Igenomix Bronchiectasis and Primary Ciliary Dyskinesia Precision Panel can be used as a diagnostic tool ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes.

Indications

The Igenomix Bronchiectasis and Primary Ciliary Dyskinesia Precision Panel is indicated in those cases where there is a clinical suspicion or imaging findings with or without the following manifestations:

- Cough and daily mucopurulent sputum production
- Blood-streaked sputum
- Shortness of breath
- Pleuritic chest pain
- Wheezing
- Fever
- Weakness

- Fatigue
- Weight loss
- Infertility
- Recurrent upper and lower respiratory tract infections
- Situs inversus (organ laterality defects)

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis and improve prognosis.
- Early initiation of treatment with a multidisciplinary team in the form of prophylactic antibiotics, chest physiotherapy, bronchodilator therapy, and adjunctive surgical resection to improve symptoms, reduce complications and control exacerbations to reduce morbidity and mortality.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

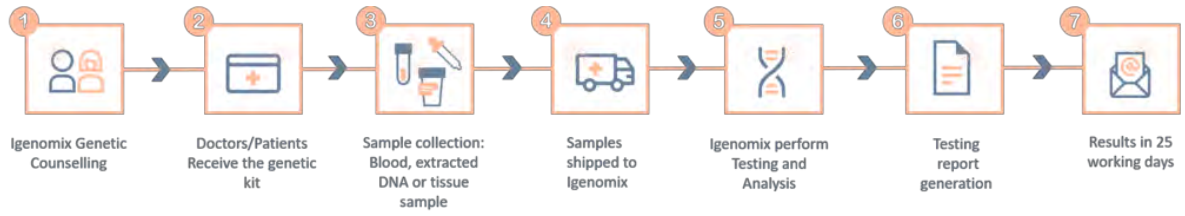
GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCA3	Idiopathic Pulmonary Fibrosis, Infant Acute Respiratory Distress Syndrome, Surfactant Metabolism Dysfunction	AR	100%	286 of 289
ARHGEF1	Immunodeficiency	AR	90.23%	2 of 2
ATM	Ataxia-telangiectasia	AD,AR	99.93%	1608 of 1632
ATP11A	Idiopathic Pulmonary Fibrosis	-	99.97%	NA of NA
B2M	Familial Visceral Amyloidosis, Hypoproteinemia	AD,AR	100%	4 of 4
BACH2	Immunodeficiency	AD	99.89%	2 of 2
BLM	Bloom Syndrome	AR	97.19%	133 of 141
BLNK	Autosomal Recessive Agammaglobulinemi	AR	97.97%	6 of 6
BTNL2	Sarcoidosis	AD	99.98%	1 of 1
CARMIL2	Immunodeficiency	AR	96.16%	NA of NA
CCDC103	Primary Ciliary Dyskinesia	AR	99.92%	6 of 6
CCDC39	Primary Ciliary Dyskinesia	AR	99.56%	48 of 52
CCDC40	Primary Ciliary Dyskinesia	AR	98%	50 of 50
CCDC65	Primary Ciliary Dyskinesia	AR	99.98%	3 of 3
CCNO	Primary Ciliary Dyskinesia	AR	99.94%	12 of 12
CD19	Common Variable Immunodeficiency	AD,AR	99.99%	7 of 7
CD79A	Autosomal Recessive Agammaglobulinemia	AR	99.99%	8 of 8
CD79B	Autosomal Recessive Agammaglobulinemia	AR	100%	3 of 3
CD81	Common Variable Immunodeficiency	AR	100%	2 of 2
CD8A	Familial CD8 Deficiency	AR	99.60%	1 of 1
CFAP221	Primary Ciliary Dyskinesia	-	89.78%	NA of NA
CFAP298	Primary Ciliary Dyskinesia	AR	na	na
CFAP300	Primary Ciliary Dyskinesia	AR	na	na
CFTR	Bronchiectasis, Congenital Bilateral Absence Of Vas Deferens, Cystic Fibrosis, Hereditary Chronic Pancreatitis, Male Infertility With Azoospermia Or Oligozoospermia	AD,AR	95.45%	1615 of 1730
CLCA4	Cystic Fibrosis	-	97.66%	NA of NA
CR2	Common Variable Immunodeficiency	AD,AR	99.92%	19 of 19
CTLA4	Autoimmune Lymphoproliferative Syndrome, Granulomatosis With Polyangiitis, Systemic Lupus Erythematosus	AD	99.97%	60 of 60
CXCR4	Whim Syndrome	AD	100%	19 of 19
DCTN4	Cystic Fibrosis	-	100%	1 of 1
DNAAF1	Primary Ciliary Dyskinesia	AR	99.55%	36 of 37
DNAAF2	Primary Ciliary Dyskinesia	AR	97.45%	7 of 8
DNAAF3	Primary Ciliary Dyskinesia	AR	98.95%	13 of 14
DNAAF4	Primary Ciliary Dyskinesia	AD,AR	99.27%	NA of NA

DNAAF5	Primary Ciliary Dyskinesia	AR	89.27%	NA of NA
DNAAF6	Primary Ciliary Dyskinesia	X,XR,G	99.63%	NA of NA
DNAH1	Primary Ciliary Dyskinesia	AR	100%	58 of 58
DNAH11	Primary Ciliary Dyskinesia	AR	99.27%	159 of 169
DNAH17	Spermatogenic Failure	AR	99.99	12 of 12
DNAH5	Primary Ciliary Dyskinesia With Or Without Situs Inversus	AR	100%	277 of 278
DNAH8	Primary Ciliary Dyskinesia	-	99.75%	12 of 12
DNAH9	Primary Ciliary Dyskinesia	AR	98.86%	19 of 19
DNAI1	Kartagener Syndrome, Primary Ciliary Dyskinesia	AR	96.91%	43 of 43
DNAI2	Primary Ciliary Dyskinesia With Or Without Situs Inversus	AR	98.89%	8 of 8
DNAJB13	Primary Ciliary Dyskinesia	AR	99.94%	3 of 3
DNAL1	Primary Ciliary Dyskinesia	AR	99.43%	5 of 5
DNMT3B	Immunodeficiency-Centromeric Instability-Facial Anomalies Syndrome	AR	100%	59 of 59
DPP9	Idiopathic Pulmonary Fibrosis	-	93.97%	1 of 1
DRC1	Primary Ciliary Dyskinesia	AR	100%	9 of 9
DSP	Idiopathic Pulmonary Fibrosis	AD,AR	99.91%	366 of 369
FAM13A	Idiopathic Pulmonary Fibrosis	-	99.91%	NA of NA
FCGR2A	Cystic Fibrosis, Systemic Lupus Erythematosus	AD,AR	93.97%	NA of NA
FOXJ1	Primary Ciliary Dyskinesia	AD	99.69%	5 of 5
GAS2L2	Primary Ciliary Dyskinesia	AR	89%	4 of 5
GAS8	Primary Ciliary Dyskinesia	AR	99.98%	6 of 6
HLA-DRB1	Diffuse Cutaneous Systemic Sclerosis, Limited Cutaneous Systemic Sclerosis, Sarcoidosis	AD,MU	97.19%	2 of 2
HYDIN	Primary Ciliary Dyskinesia	AR	81.70%	45 of 63
ICOS	Common Variable Immunodeficiency	AD,AR	100%	4 of 5
IGHM	Autosomal Recessive Agammaglobulinemia	AR	100%	NA of NA
IGLL1	Autosomal Recessive Agammaglobulinemia	AR	100%	2 of 2
IL21R	IL21R Immunodeficiency	AR	99.97%	10 of 10
IL6ST	Hyper-IgE Recurrent Infection Syndrome	AR	99.34%	2 of 2
IRF8	Immunodeficiency 32A, Immunodeficiency 32B, Mendelian Susceptibility To Mycobacterial Diseases Due To Partial IRF8 Deficiency	AD,AR	100%	9 of 9
IRF9	Immunodeficiency, Susceptibility To Viral Infections	AR	100%	5 of 5
LRBA	Common Variable Immunodeficiency	AR	99.91%	79 of 81
LRRC56	Primary Ciliary Dyskinesia	AR	99.77%	5 of 5
LRRC6	Primary Ciliary Dyskinesia	AR	99.88%	21 of 21
LRRC8A	Autosomal Dominant Agammaglobulinemia	AD	100%	2 of 2
MCIDAS	Primary Ciliary Dyskinesia	AR	99.92%	4 of 4
MS4A1	Common Variable Immunodeficiency	AR	100%	2 of 2
MUC5B	Idiopathic Pulmonary Fibrosis	AD	99.89%	12 of 12
NBN	Nijmegen Breakage Syndrome	AR,MU,P	100%	200 of 200
NCKAP1L	Immunodeficiency With Autoinflammation	AR	100%	NA of NA
NEK10	Primary Ciliary Dyskinesia	AR	99.95%	3 of 3
NFKB1	Common Variable Immunodeficiency	AD	99.98%	38 of 41
NFKB2	Common Variable Immunodeficiency	AD	100%	22 of 22
NME8	Primary Ciliary Dyskinesia	AR	99.99%	9 of 9
ODAD1	Primary Ciliary Dyskinesia	AR	99.68%	10 of 10
ODAD2	Primary Ciliary Dyskinesia	AR	97.30%	26 of 28
ODAD3	Primary Ciliary Dyskinesia	AR	95%	4 of 4
ODAD4	Primary Ciliary Dyskinesia	AR	na	na
OFD1	Primary Ciliary Dyskinesia	X,XR,XD,G	98.09%	NA of NA
PARN	Idiopathic Pulmonary Fibrosis, Pulmonary Fibrosis And/Or Bone Marrow Failure, Telomere-Related	AD,AR	99.98%	33 of 33
PGM3	Immunodeficiency	AR	99.99%	17 of 17
PIK3CD	Combined Immunodeficiency With Facioculoskeletal Anomalies	AD	100%	23 of 23
PIK3R1	Autosomal Recessive Agammaglobulinemia, Immunodeficiency	AD,AR	99.89%	29 of 29
POLD1	Mandibular Hypoplasia, Deafness, Progeroid Features, And Lipodystrophy Syndrome	AD	100%	40 of 41
PRKCD	Autoimmune Lymphoproliferative Syndrome, Common Variable Immunodeficiency	AR	100%	9 of 9
RAC2	Immunodeficiency With Defective Neutrophil Chemotaxis And Lymphopenia, Neutrophil Immunodeficiency Syndrome	AD,AR	100%	5 of 5

RASGRP1	Autoimmune Lymphoproliferative Syndrome, Immunodeficiency	AR	98.41%	8 of 9
RIN2	Macrocephaly, Alopecia, Cutis Laxa, And Scoliosis, RIN2 Syndrome	AR	99.60%	4 of 4
RIPK1	Autoinflammation With Episodic Fever And Lymphadenopathy, Immunodeficiency	AD,AR	98.03%	12 of 14
RPGR	Primary Ciliary Dyskinesia, X-linked Retinitis Pigmentosa And Sinorespiratory Infections, Withor Without Deafness	X,XR,G	94%	NA of NA
RSPH1	Primary Ciliary Dyskinesia	AR	100%	10 of 10
RSPH3	Primary Ciliary Dyskinesia	AR	99.85%	5 of 5
RSPH4A	Primary Ciliary Dyskinesia	AR	99.98%	27 of 27
RSPH9	Primary Ciliary Dyskinesia	AR	100%	13 of 13
RTEL1	Dyskeratosis Congenita, Idiopathic Pulmonary Fibrosis, Pulmonary Fibrosis And/Or Bone Marrow Failure, Telomere-Related	AD,AR	99.73%	127 of 131
SCNN1A	Bronchiectasis With Or Without Elevated Sweat Chloride, Idiopathic Bronchiectasis	AD,AR	99.95%	46 of 46
SCNN1B	Idiopathic Bronchiectasis	AD,AR	100%	56 of 56
SCNN1G	Bronchiectasis With Or Without Elevated Sweat Chloride, Idiopathic Bronchiectasis	AD,AR	100%	28 of 28
SFTPA1	Idiopathic Pulmonary Fibrosis		100%	4 of 4
SFTPA2	Idiopathic Pulmonary Fibrosis	AD	99.98%	6 of 6
SFTPC	Idiopathic Pulmonary Fibrosis, Infant Acute Respiratory Distress Syndrome, Pulmonary Fibrosis, Surfactant Metabolism Dysfunction	AD	99.84%	83 of 83
SLC29A3	Histiocytosis-Lymphadenopathy Plus Syndrome	AR	100%	32 of 32
SPAG1	Primary Ciliary Dyskinesia	AR	94.80%	11 of 12
SPEF2	Primary Ciliary Dyskinesia	AR	99.60%	10 of 13
STAT1	Autoimmune Enteropathy And Endocrinopathy-Susceptibility To Chronic Infections Syndrome, Immunodeficiency, Mycobacterial And Viral Infections	AD,AR	100%	138 of 138
STK36	Primary Ciliary Dyskinesia	-	100%	5 of 5
STN1	Idiopathic Pulmonary Fibrosis	AR	99.87%	NA of NA
STX1A	Cystic Fibrosis	-	97%	3 of 3
TAP1	Bare Lymphocyte Syndrome, Type I	AR	100%	7 of 7
TAP2	Bare Lymphocyte Syndrome, Type I	AR	100%	9 of 9
TAPBP	Bare Lymphocyte Syndrome, Type I	AR	93.99%	1 of 1
TCF3	Autosomal Dominant Agammaglobulinemia	AD	99.98%	7 of 7
TERC	Dyskeratosis Congenita, Idiopathic Pulmonary Fibrosis, Pulmonary Fibrosis And/Or Bone Marrow Failure, Telomere-Related	AD	na	na
TERT	Dyskeratosis Congenita, Idiopathic Pulmonary Fibrosis, Pulmonary Fibrosis And/Or Bone Marrow Failure, Telomere-Related	AD,AR	99.09%	194 of 197
TGFB1	Cystic Fibrosis, Immunodeficiency And Encephalopathy	AD,AR	99.75%	24 of 24
TNFRSF13B	Common Variable Immunodeficiency	AD,AR	100%	50 of 50
TNFRSF13C	Common Variable Immunodeficiency	AD,AR	99.20%	3 of 3
TNFSF12	Common Variable Immunodeficiency	-	95.06%	1 of 1
TTC12	Primary Ciliary Dyskinesia	AR	99.97%	NA of NA
WDR1	Periodic Fever, Immunodeficiency, And Thrombocytopenia Syndrome	AR	100%	9 of 9
ZMYND10	Primary Ciliary Dyskinesia	AR	99.98%	16 of 16
ZNF341	Autosomal Recessive Hyper-IgE Recurrent Infection Syndrome	AR	100%	6 of 6

* Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial
** HGMD: Number of clinically relevant mutations according to HGMD

Methodology



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- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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