Greenwood Diagnostic Labs

Pulmonary Disease

Genetic Testing

Genetic Testing for Pulmonary Diseases

Condition	Gene	Central Hypoventilation Sequencing Panel	Dyskeratosis Congenita Sequencing Panel	Hermansky-Pudlak and Pulmonary Fibrosis Sequencing Panel	Primary Ciliary Dyskinesia & Cystic Fibrosis Sequencing Panel	Pulmonary Arterial Hypertension Sequencing Panel	Surfactant Dysfunction & Respiratory Distress in Premature Infants Sequencing Panel	Comprehensive Pulmonary Sequencing Panel
Alström Syndrome	*ALMS1*							x
Alveolar Capillary Dysplasia with misalignment of pulmonary veins	*FOXF1*						x	x
Atrial Fibrillation, familial, 7	*KCNA5*					x		x
Autoimmune Disease, multisystem, infantile-onset, 1	*STAT3*							x
Autoimmune Interstitial Lung, Joint, and Kidney Disease	*COPA*			x				x
Bronchiectasis with or without elevated sweat chloride 1 Pseudohypoaldosteronism, type I	*SCNN1B*				x			x
Bronchiectasis with or without elevated sweat chloride 2 Pseudohypoaldosteronism, type I	*SCNN1A*				x			x
Bronchiectasis with or without elevated sweat chloride 3 Liddle Syndrome Pseudohypoaldosteronism, type I	*SCNN1G*				x			x
Cerebroretinal Microangiopathy with calcifications and cysts	*CTC1*		x	x				x
Central Hypoventilation Syndrome	*MYO1H*	x						x
Central Hypoventilation Syndrome, congenital	*ASCL1*	x						x
Central Hypoventilation Syndrome, congenital, with or without Hirschprung Disease	*PHOX2B*	x						x
Choreoathetosis, Hypothyroidism, and Neonatal Respiratory Distress	*NKX2-1*			x			x	x
Chronic Obstructive Pulmonary Disease (COPD), susceptibility to	*SFTPD*			x				x
Ciliary Dyskinesia, primary, 1, with or without Situs Inversus	*DNAI1*				x			x
Ciliary Dyskinesia, primary, 2	*DNAAF3*				x			x
Ciliary Dyskinesia, primary, 3, with or without Situs Inversus	*DNAH5*				x			x
Ciliary Dyskinesia, primary, 6	*NME8*				x			x
Ciliary Dyskinesia, primary, 7, with or without Situs Inversus	*DNAH11*				x			x
Ciliary Dyskinesia, primary, 9, with or without Situs Inversus	*DNAI2*				x			x
Ciliary Dyskinesia, primary, 10	*DNAAF2*				x			x
Ciliary Dyskinesia, primary, 11	*RSPH4A*				x			x
Ciliary Dyskinesia, primary, 12	*RSPH9*				x			x
Ciliary Dyskinesia, primary, 13	*DNAAF1*				x			x
Ciliary Dyskinesia, primary, 14	*CCDC39*				x			x
Ciliary Dyskinesia, primary, 15	*CCDC40*				x			x
Ciliary Dyskinesia, primary, 16	*DNAL1*				x			x
Ciliary Dyskinesia, primary, 17	*CCDC103*				x			x
Ciliary Dyskinesia, primary, 18	*DNAAF5*				x			x
Ciliary Dyskinesia, primary, 19	*LRRC6*				x			x
Ciliary Dyskinesia, primary, 20	*CCDC114*				x			x
Ciliary Dyskinesia, primary, 21	*DRC1*				x			x
Ciliary Dyskinesia, primary, 22	*ZMYND10*				x			x
Ciliary Dyskinesia, primary, 23	*ARMC4*				x			x
Ciliary Dyskinesia, primary, 24	*RSPH1*				x			x
Ciliary Dyskinesia, primary, 25	*DYX1C1*				x			x
Ciliary Dyskinesia, primary, 26	*C21ORF59*				x			x
Ciliary Dyskinesia, primary, 27	*CCDC65*				x			x
Ciliary Dyskinesia, primary, 28	*SPAG1*				x			x
Ciliary Dyskinesia, primary, 29	*CCNO*				x			x

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Ciliary Dyskinesia, primary, 30	*CCDC151*				x			x
Ciliary Dyskinesia, primary, 32	*RSPH3*				x			x
Ciliary Dyskinesia, primary, 33	*GAS8*				x			x
Ciliary Dyskinesia, primary, 34	*DNAJB13*				x			x
Ciliary Dyskinesia, primary, 35	*TTC25*				x			x
Ciliary Dyskinesia, primary, candidate gene	*DNAH8*				x			x
Ciliary Dyskinesia, primary, candidate gene multiple morphological abnormalities of the sperm flagella	*DNAH1*				x			x
Corneal Dystrophy, posterior polymorphous	*GRHL2*		x	x				x
Costello Syndrome	*HRAS*							x
Cutis Laxa Supravalvar Aortic Stenosis	*ELN*							x
Cutis Laxa, autosomal dominant 2 Cutis Laxa, autosomal recessive, type IA	*FBLN5*							x
Cutis Laxa, autosomal recessive, type IB	*EFEMP2*							x
Cutis Laxa, autosomal recessive, type IC	*LTBP4*							x
Cystic Fibrosis	*CFTR*				x			x
Dyskeratosis Congenita, autosomal dominant 1 Pulmonary Fibrosis, idiopathic, susceptibility to	*TERC*		x	x				x
Dyskeratosis Congenita, autosomal recessive 1	*NOP10*		x	x				x
Dyskeratosis Congenita, autosomal recessive 2	*NHP2*		x	x				x
Dyskeratosis Congenita, autosomal dominant 2 Dyskeratosis Congenita, autosomal recessive 4 Pulmonary Fibrosis and/or bone marrow failure, telomere-related, 1	*TERT*		x	x				x
Dyskeratosis Congenita, autosomal dominant 3	*TINF2*		x	x				x
Dyskeratosis Congenita, autosomal recessive 3	*WRAP53*		x	x				x
Dyskeratosis Congenita, autosomal dominant 4 Dyskeratosis Congenita, autosomal recessive 5 Pulmonary Fibrosis and/or bone marrow failure, telomere-related, 3	*RTEL1*		x	x				x
Dyskeratosis Congenita, autosomal dominant 6 Dyskeratosis Congenita, autosomal recessive 7	*ACD*		x	x				x
Dyskeratosis Congenita, autosomal recessive 6 Pulmonary Fibrosis and/or bone marrow failure, telomere-related, 4	*PARN*		x	x				x
Dyskeratosis Congenita, X-linked	*DKC1*		x	x				x
Emphysema due to AAT Deficiency Emphysema-cirrhosis, due to AAT Deficiency Hemorrhagic Diathesis due to Antithrombin Pittsburgh	*SERPINA1*							x
Gaucher Disease, Type I	*GBA*							x
Glycogen Storage Disease II, Pompe Disease	*GAA*							x
Hermansky-Pudlak Syndrome 1	*HPS1*			x				x
Hermansky-Pudlak Syndrome 2	*AP3B1*			x				x
Hermansky-Pudlak Syndrome 3	*HPS3*			x				x
Hermansky-Pudlak Syndrome 4	*HPS4*			x				x
Hermansky-Pudlak Syndrome 5	*HPS5*			x				x
Hermansky-Pudlak Syndrome 6	*HPS6*			x				x
Hermansky-Pudlak Syndrome 7	*DTNBP1*			x				x
Hermansky-Pudlak Syndrome 8	*BLOC1S3*			x				x
Hermansky-Pudlak Syndrome 9	*BLOC1S6*			x				x

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Hermansky-Pudlak Syndrome 10	*AP3D1*			x				x
Hereditary Hemorrhagic Telangiectasia Syndrome Juvenile Polyposis	*SMAD4*					x		x
Hyper-IgE Recurrent Infection Syndrome	*DOCK8*							x
Intellectual Disability, autosomal dominant 31	*PURA*							x
Interstitial Lung & Liver Disease	*MARS*			x			x	x
Ischiocoxopodopatellar Syndrome with or without Pulmonary Arterial Hypertension	*TBX4*					x		x
LIG4 Syndrome	*LIG4*		x	x				x
Lymphangioleiomyomatosis Tuberous Sclerosis-1	*TSC1*							x
Lymphangioleiomyomatosis, somatic Tuberous Sclerosis-2	*TSC2*							x
Lysinuric Protein Intolerance	*SLC7A7*						x	x
Marfan Syndrome Geleophysic Dysplasia, 2	*FBN1*					x		x
Mucociliary Clearance Disorder	*MCIDAS*				x			x
Nephronophthisis 2, infantile	*INVS*				x			x
Neurofibromatosis, type 1 Neurofibromatosis-Noonan Syndrome	*NF1*					x		x
Pneumothorax, primary spontaneous Birt-Hogg-Dubé Syndrome	*FLCN*							x
Poikiloderma, hereditary fibrosing, with tendon contractures, Myopathy, and Pulmonary Fibrosis	*FAM111B*			x				x
Poikiloderma with Neutropenia	*USB1*		x	x				x
Prolidase Deficiency	*PEPD*							x
Pulmonary Arterial Hypertension	*ATP13A3*					x		x
Pulmonary Arterial Hypertension	*KLF2*					x		x
Pulmonary Arterial Hypertension	*SMAD1*					x		x
Pulmonary Arterial Hypertension	*AQP1*					x		x
Pulmonary Fibrosis	*NAF1*			x				x
Pulmonary Fibrosis, idiopathic	*SFTPA2*			x				x
Pulmonary Fibrosis, idiopathic, candidate gene	*ELMOD2*			x				x
Pulmonary Fibrosis, idiopathic, susceptibility to	*MUC5B*			x				x
Pulmonary Fibrosis, idiopathic, susceptibility to	*SFTPA1*			x				x
Pulmonary Arterial Hypertension, childhood idiopathic	*BMPR1B*					x		x
Pulmonary Hypertension, familial primary, 1, with or without HHT Pulmonary Hypertension, primary, fenfluramine or dexfenfluramine-associated Pulmonary venoocclusive disease 1	*BMPR2*					x		x
Pulmonary Hypertension, primary, 2	*SMAD9*					x		x
Pulmonary Hypertension, primary, 3	*CAV1*					x		x
Pulmonary Hypertension, primary, 4	*KCNK3*					x		x
Pulmonary Venoocclusive Disease 2	*EIF2AK4*					x		x
Rajab Interstital Lung Disease with brain calcifications	*FARSB*			x		x		x
Retinitis pigmentosa, X-linked, and Sinorespiratory Infections, with or without deafness	*RPGR*				x			x
Respiratory failure due to altered lung development and sometimes accompanying Pulmonary Hypertension	*FLNA*					x	x	x

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Simpson-Golabi-Behmel Syndrome, type 2 Oral-Facio-Digital Syndrome I	*OFD1*					x		x
STING-Associated Vasculopathy, infantile onset	*TMEM173*			x			x	x
Stromme Syndrome	*CENPF*					x		x
Surfactant Metabolism Dysfunction, pulmonary, 1	*SFTPB*			x			x	x
Surfactant Metabolism Dysfunction, pulmonary, 2	*SFTPC*			x			x	x
Surfactant Metabolism Dysfunction, pulmonary, 3	*ABCA3*			x	x		x	x
Surfactant Metabolism Dysfunction, pulmonary, 4	*CSF2RA*			x			x	x
Surfactant Metabolism Dysfunction, pulmonary, 5	*CSF2RB*						x	x
Telangiectasia, hereditary hemorrhagic, type 1	*ENG*				x			x
Telangiectasia, hereditary hemorrhagic, type 2	*ACVRL1*				x			x
Telangiectasia, hereditary hemorrhagic, type 5	*GDF2*				x			x
Vesicoureteral Reflux 3	*SOX17*				x			x

* targeted analysis of only the c.-3133G>T variant within the promoter region of the *MUC5B* gene

Cystic Fibrosis (CFTR) Genetic Testing

	Sequencing	Del/Dup	Targeted Analysis	Targeted Analysis (Prenatal)
CFTR	81223	81222	81221	81221
TAT	4 Weeks	4 Weeks	2 Weeks	2 Weeks
Price	$1,500	$700	$350	$1,000

NGS Pulmonary Panels

	Comprehensive Pulmonary Panel	Central Hypoventilation Sequencing Panel	Dyskeratosis Congenita Sequencing Panel	Hermansky-Pudlak and Pulmonary Fibrosis Sequencing Panel	Primary Ciliary Dyskinesia and Cystic Fibrosis Sequencing Panel	Pulmonary Arterial Hypertension Sequencing Panel	Surfactant Dysfunction and Respiratory Distress in Premature Infants Sequencing Panel
Genes	124 Genes	3 Genes	14 Genes	40 Genes	42 Genes	22 Genes	11 Genes
CPT Code	81479	81479	81479	81479	81479	81479	81479
TAT	8-10 Weeks	8-10 Weeks	8-10 Weeks	8-10 Weeks	8-10 Weeks	8-10 Weeks	8-10 Weeks
Price	$3,500	$2,000	$2,500	$3,000	$3,000	$3,000	$2,500

Greenwood Diagnostic Labs


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Genetic testing can play a significant role in the diagnosis and management of pulmonary disorders. A specific diagnosis may provide prognostic insight, guide treatment, prevent additional and unnecessary testing, and give valuable recurrence risk information.

Greenwood Diagnostic Labs offer a Comprehensive Pulmonary Panel and six subpanels for more specific phenotypes. These next generation sequencing panels offer an efficient and thorough molecular analysis for the heterogeneous and complex conditions associated with hereditary pulmonary disorders.

Contact a laboratory genetic counselor

+1 (800) 473-9411

Phones are answered Monday-Friday from 8am-5pm.

Specimen requirements

4-5 ml of peripheral blood should be collected in EDTA (lavender top) vacutainer tube.

(Saliva or extracted DNA will also be accepted.)

Shipping requirements

Ship at room temperature to:

106 Gregor Mendel Circle
Greenwood, SC 29646

Blood and Saliva Sample Kits

Available by request

106 Gregor Mendel Circle
Greenwood, SC 29646

Phone: +1 (800) 473-9411
Fax: + 1 (864) 941-8141

GGC.org/testfinder

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