



# PULMONOLOGY TESTING



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## GENETIC TESTING FOR PULMONOLOGY

Inherited pulmonary diseases are a group of lung disorders with variable clinical presentation and frequently have significant phenotypic overlap. The diseases can affect the airways (e.g. cystic fibrosis and primary ciliary dyskinesia), parenchyma (pulmonary fibrosis, Birt Hogg Dube syndrome and tuberous sclerosis) and vasculature of the lung. Rare lung diseases generally affect individuals from birth through about age 60. They are in many cases serious, chronic, and can be devastating. Once properly diagnosed, they often require expensive, long-term treatments. Genetic diagnostics by NGS offers a rapid approach to correct clinical diagnosis and early and personalized intervention.



### What genetic diagnostics can offer patients with pulmonological diseases ?

Genetic diagnostics are often the most efficient way to subtype hereditary pulmonary diseases, and they provide the necessary information to make confident individualized treatment and management decisions. Specifically, variation in the CFTR gene that causes cystic fibrosis, is targeted in this category.

In addition to cystic fibrosis, determining the exact underlying genetic defect in any hereditary pulmonary disease heavily affects genetic counseling and risk assessment.

Identifying at-risk family members makes it possible to begin preventive treatments and/or make lifestyle recommendations. It also justifies routine follow-ups by health care professionals. Genetic diagnostics can help in family planning.

## TEST PERFORMANCE

Elite Clinical Laboratory offer a Comprehensive Pulmonology Panel. It is a 68 gene panel that includes assessment of non-coding variants. The genes on the panel have been carefully selected based on scientific literature, mutation databases and our experience.

## CLINICAL INTERPRETATION

We provide customers with the most comprehensive clinical report available on the market. Clinical interpretation requires a fundamental understanding of clinical genetics and genetic principles. At Elite Clinical Laboratory, our PhD molecular geneticists, medical geneticists and clinical consultants prepare the clinical statement together by evaluating the identified variants in the context of the phenotypic information provided in the requisition form. Our goal is to provide clinically meaningful statements that are understandable for all medical professionals regardless of whether they have formal training in genetics.



**Panel Gene List:** ABCA3, CCDC39, CCDC40, CFTR, CHAT, CHRNA1, CHRN1, CHRND, CHRNE, COLQ, CSF2RA, CSF2RB, DKC1, DNAAF1, DNAAF2, DNAH1, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, EDN3, EFEMP2, ELMOD2, ELN, FBLN5, FLCN, FOXF1, GAS8, GLRA1, HPS1, HPS4, ITGA3, LTBP4, MECP2, NAF1, NF1, NKX2-1, NME8, PARN, PHOX2B, PIH1D3#, RAPSN, RET, RSPH3, RSPH4A, RSPH9, RTEL1, SCN4A, SCNN1A, SCNN1B, SERPINA1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC6A5, SLC7A7, SMPD1, STAT3, TERC, TERT, TINF2, TSC1, TSC2, ZEB2

### Specimen Requirements:

Buccal Swab (Wet/Dry) or Extracted DNA (5ug)

### Turnaround Time:

2-3 weeks

# "Dedicated towards betterment"

Elite Clinical Laboratory is a full service, national diagnostic testing laboratory headquartered in Houston, Texas with concentrations in clinical diagnostics, toxicology, genetic sequencing and molecular testing. Elite Clinical Laboratory is devoted to redefining diagnostic services by providing medical practitioners and their patients with exceptional customer service paired with the most advanced and informative medical analytics to assist them in making effective treatment decisions.

Elite Clinical Laboratory fully automated laboratory utilizes state-of-the-art technologies to deliver high quality test results and service while exceeding the turnaround time requirements and demands of our physician clients. Elite Clinical Laboratory currently analyzes samples for hundreds of thousands of patients per year from providers and healthcare facilities all across the nation.

As our clients have trusted our laboratory with being an analytical and integral part of their patients' diagnosis and treatment process, we believe in respecting that trust with continuous dedication to customer satisfaction and support. We join our clients and physicians in their belief that patient care is and always will be the number one priority. Elite's personalized support and professional service continues to exceed the expectations of our valued clients, providers and facilities. More healthcare facilities and providers, in private practices, in hospitals and in long term care facilities, are placing their trust in Elite Clinical Laboratory; and, together we are transforming advanced diagnostic information into knowledge and superior treatment options for more and more patients every day.

