

# A Review on Cystic Fibrosis Disease

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**Abstract:** Cystic fibrosis is a monogenic disease considered to affect at least 100 000 people worldwide. Mutations in CFTR, the gene encoding the epithelial ion channel that normally transports chloride and bicarbonate, lead to impaired mucus hydration and clearance. Classical cystic fibrosis is thus characterised by chronic pulmonary infection and inflammation, pancreatic exocrine insufficiency, male infertility, and might include several comorbidities such as cystic fibrosis-related diabetes or cystic fibrosis liver disease. This autosomal recessive disease is diagnosed in many regions following newborn screening, whereas in other regions, diagnosis is based on a group of recognised multiorgan clinical manifestations, raised sweat chloride concentrations, or CFTR mutations. Disease that is less easily diagnosed, and in some cases affecting only one organ, can be seen in the context of gene variants leading to residual protein function. Management strategies, including augmenting mucociliary clearance and aggressively treating infections, have gradually improved life expectancy for people with cystic fibrosis. However, restoration of CFTR function via new small molecule modulator drugs is transforming the disease for many patients. Clinical trial pipelines are actively exploring many other approaches, which will be increasingly needed as survival improves and as the population of adults with cystic fibrosis increases. Here, we present the current understanding of CFTR mutations, protein function, and disease pathophysiology, consider strengths and limitations of current management strategies, and look to the future of multidisciplinary care for those with cystic fibrosis.

**Keywords:** Cystic fibrosis