

*Commentary*

# Cystic fibrosis: Causes, symptoms, diagnosis, and advances in treatment

**Brinnlie Peterson\***

Department of Dermatology, University of Utah, Salt Lake City, USA.

Received: 05-Jun-2023, Manuscript No. IJMMS-23-104805; Editor assigned: 08-Jun-2023, PreQC No. IJMMS-23-104805 (PQ); Reviewed: 23-Jun-2023, QC No. IJMMS-23-104805; Revised: 30-Jun-2023, Manuscript No. IJMMS-23-104805 (R); Published: 07-Jul-2023

**DESCRIPTION**

Cystic fibrosis (CF) is a chronic and complex genetic disorder that impacts multiple organ systems, most notably the lungs and digestive system. This article provides a comprehensive overview of cystic fibrosis, covering its causes, symptoms, diagnosis, and available treatment options. It also emphasizes the ongoing efforts to improve the lives of individuals living with this challenging disease (Afzal et al., 1991).

**Causes and genetics of cystic fibrosis**

Cystic fibrosis is caused by a mutation in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene. The *CFTR* gene is responsible for producing a protein that regulates the movement of salt and water across cell membranes (Hess et al., 1998). When the *CFTR* gene is mutated, it results in a defective *CFTR* protein, disrupting the normal balance of salt and water in the body. Consequently, this leads to the production of thick, sticky mucus in various organs, particularly the lungs and digestive system (Tang et al., 2022).

The inheritance pattern of cystic fibrosis is autosomal recessive. This means that both parents must carry a mutated *CFTR* gene for a child to develop the condition. If both parents are carriers, each pregnancy carries a 25% chance of the child being born with cystic fibrosis. It is essential for carriers to undergo genetic counseling to fully understand the risks associated with having a child with CF (Wei et al., 2019).

**Symptoms and complications of cystic fibrosis**

The symptoms of cystic fibrosis can vary from person to person, but they primarily affect the respiratory and digestive systems. Common symptoms include

- Persistent cough with thick mucus
- Frequent lung infections, such as pneumonia or

bronchitis

- Shortness of breath and wheezing
- Poor growth and weight gain despite a good appetite
- Foul-smelling, bulky stools
- Difficulty absorbing nutrients, leading to malnutrition and vitamin deficiencies
- Nasal polyps and sinus infections

Over time, the accumulation of thick mucus can lead to progressive lung damage and respiratory complications. Individuals with cystic fibrosis are also at a higher risk of developing other conditions such as diabetes, liver disease, osteoporosis, and infertility (Wang et al., 2021). It is crucial for individuals with CF to receive regular medical care to monitor and manage these potential complications effectively.

**Diagnosis of cystic fibrosis**

Cystic fibrosis is typically diagnosed through a combination of screening tests, clinical evaluation, and genetic testing. Newborn screening programs are available in many countries, allowing for early detection of CF. These screening tests involve collecting a small blood sample or using a special swab to collect sweat. The sweat chloride test, which measures the amount of salt in sweat, is a commonly used diagnostic tool (Ye et al., 2016).

Genetic testing plays a vital role in confirming the presence of *CFTR* gene mutations. It helps identify the specific genetic variations responsible for cystic fibrosis. Understanding the genetic makeup of an individual with CF can provide valuable information for personalized treatment approaches.

**Treatment options for cystic fibrosis**

While there is currently no cure for cystic fibrosis, advancements in medical care have significantly improved the prognosis and quality of life for individuals with the condition. Treatment approaches for CF aim to manage symptoms, prevent

\*Corresponding author. Brinnlie Peterson, E-mail: [b\\_peterson@yahoo.fr](mailto:b_peterson@yahoo.fr)

complications, and improve overall health. Some key treatment options include

**Airway clearance techniques:** Regular use of techniques such as chest physiotherapy, positive expiratory pressure devices, and exercise can help loosen and clear mucus from the lungs.

**Medications:** Inhalers, antibiotics, and mucus-thinning medications are used to control respiratory symptoms and prevent infections.

**Nutritional support:** A high-calorie diet, pancreatic enzyme replacement therapy, and vitamin supplements are prescribed to ensure adequate nutrition and prevent malabsorption.

**Gene-targeted therapies:** Newer medications called CFTR modulators are designed to target specific *CFTR* gene mutations and improve CFTR protein function in select patients.

**Lung transplantation:** In severe cases where lung function significantly deteriorates, lung transplantation may be considered as a treatment option.

**Ongoing research and future perspectives:** Researchers and scientists are actively exploring new therapeutic approaches and treatment strategies for cystic fibrosis. Advances in gene therapy, precision medicine, and targeted therapies offer hope for improved outcomes and disease management. Gene editing techniques, such as CRISPR-Cas9, hold promise for correcting the underlying genetic defects in cystic fibrosis. These advancements have the potential to provide more effective and individualized treatments for individuals with CF.

Furthermore, efforts are underway to enhance supportive care, mental health support, and multidisciplinary care models to address the complex needs of individuals with cystic fibrosis. This comprehensive approach acknowledges that managing CF involves not only medical interventions but also psychological and social support.

## CONCLUSION

Cystic fibrosis is a complex genetic disorder that affects multiple organ systems, primarily the lungs and digestive

system. Although it presents significant challenges, advances in medical care and ongoing research have greatly improved the prognosis and quality of life for individuals living with CF. With continued research, collaborative efforts, and advancements in therapeutic approaches, the future holds promise for further improvements in the treatment and management of cystic fibrosis. These developments aim to enhance the lives of those affected by this complex disease and provide hope for a brighter future.

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