CASE REPORT

DOI: 10.4274/ejgg.galenos.2025.2025-2-1
Eur J Geriatr Gerontol 2025;7(2):59-61

Caring for Adult-onset Cystic Fibrosis Diagnosed at the Age of 79: A Case Report

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Abstract

We present a case of cystic fibrosis (CF) diagnosed at the age of 79, one of the oldest CF cases known to date. Our patient had an unconventional CF genotype, leading to an odyssey of a clinical journey that involved frequent treatment modifications and advanced genetic testing. The course was complicated by multiple prolonged recurrent hospitalizations, requiring extensive geriatrics, and pulmonary multidisciplinary collaborative care. This case illustrates the complexity of caring for geriatric patients with CF, highlighting the following key geriatrics M's: (m)ulti-complexity, (m)obility, (m)edications, and what (m)atters most.

Keywords: Adult-onset cystic fibrosis, geriatrics, matters most, multi-complexity, targeted therapies

Introduction

Cystic fibrosis (CF) is a monogenic disorder caused by mutations of the CF transmembrane *conductance regulator (CFTR)* gene encoding an ion channel, characterized by pulmonary infection and other multi-organ dysfunctions (1) associated with thick secretions due to impaired chloride transport. Recently, gene therapy has made a breakthrough in CF treatment. CF, has been rarely reported in older patients during bronchiectasis workup. This case of late-onset CF illustrates the necessity of an appropriate geriatrics approach according to the changing landscape of genetic disorders in older adults.

Case Report

An 84-year-old female presented to the outpatient geriatrics clinic to establish care for chronic shortness of breath and debility after hospitalization due to pneumonia. Her medical history included advanced adult-onset CF; bronchiectasis; multiple infections, including multidrug-resistant *Pseudomonas aeruginosa* and *Stenotrophomonas maltophilia* pneumonia; hypoxic respiratory failure. She had frequent CF exacerbations (6-8 per year), requiring prolonged hospitalizations and home oxygen therapy. Her health was complicated by severe deconditioning, complex polypharmacy with chronic antibiotic suppressive therapy, and iatrogenic pancytopenia.

Our patient was diagnosed with CF at the age of 79, at an outside health system in the setting of progressive bronchiectasis with recurrent pulmonary infections over 15 years. She was previously free of sinopulmonary symptoms. She had no risk factors for lung disease and no family history of genetic disorders. Diagnosis was established via serial sweat tests (70 mmol/L and 63 mmol/L in the following year) and clinical criteria. *CFTR* gene sequencing and deletion/duplication testing at the time of diagnosis showed homozygous variants of the 11TG/7T alleles, which are not the common pathogenic variants. A repeat nasal swab was sent to an outside research-based multi-analysis program and returned with the same genetic profile.

The patient lived with her husband, who was her primary caregiver. The patient encountered significant challenges

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Received: 06.02.2025 **Accepted:** 29.05.2025 **Epub:** 07.07.2025

Cite this article as: Lee J, Gorenchtein M. Caring for adult-onset cystic fibrosis diagnosed at the age of 79: a case report. Eur J Geriatr Gerontol. [Epub Ahead of Print]

following the CF treatment regimen, which entailed variable daily and weekly multi-drug antibiotic dosing, along with medications for symptomatic management, such as dornase alfa. Hopeful for treatment optimization, the patient underwent multiple bronchoscopies for respiratory fluid sampling. This was part of the *in vitro* research program at an outside institution the cellular response to novel CF-targeted therapies, which yielded negative results. She opted for port-a-cath placement for chronic antibiotic suppressive therapy.

Throughout the patient's remaining clinical course, the geriatrics team provided coordination of care, including consolidating the treatment regimen and educating the patient on CF complications. Geriatrics maintained frequent communication with the pulmonary team, especially to facilitate CF medication changes, follow-up investigations, and coordination of supportive services (such as physical therapy, occupational therapy, skilled nursing, and transportation). Geriatric specialists coordinated regular multidisciplinary meetings to discuss treatment goals, prognosis, realistic expectations, hospice, and quality of life, to find a balance between disease treatment and what matters most. Geriatrics also provided co-management and transition of care during each hospitalization. As the patient's functional status continued to decline with increasing caregiver burden, geriatrics facilitated the transition to a home visiting geriatric program. The patient subsequently had another hospitalization for CF exacerbation, during which she was transitioned to inpatient hospice with terminal extubation, and she passed shortly afterward.

Consent for Publication

Informed consent for the publication of this case report and any associated clinical details was obtained from the patient's next of kin (her husband, the primary caregiver) and documented in the Electronic Medical Record.

Discussion

CF is the most common autosomal recessive hereditary disorder in the Caucasian population (2). The condition is diagnosed with either the sweat chloride test coupled with characteristic CF phenotype or the identification of two pathogenic variants of the *CFTR* gene. Different genetic mutations define the mechanism of CFTR protein dysfunction (Class I-VI). Two main types of CFTR modulator therapy are available: 1) potentiators, which are small molecules aiding in opening the chloride channel leading to CFTR activation, 2) correctors, which aid in repairing the CFTR trafficking defects (3). Novel target treatments (modulators) have recently been approved for selected genotypes, such as Phe508del and Gly551Asp. Since many mutations have multiple defects with overlapping mechanisms, a significant amount of work has been geared toward developing novel targeted combinatorial therapeutics (3,4). The triple combination CFTR modulator drug, ivacaftor/tezacaftor/elexacaftor (Trikafta), including one potentiator and two correctors, has demonstrated superior efficacy for a greater number of CF patients (5).

There are about 2,000 known pathogenic variants in CF, demonstrating genetic heterogeneity. For example, extensive genetic and functional analysis of CFTR in CF patients has identified key regions with disruptions in the polymorphic TG and T tracts (6,7). These molecular lesions are associated with altering the physiological splicing of the *CFTR* gene, resulting in loss of function in the CFTR protein (6), suggesting an important role in CF pathogenesis. Additional research including molecular and functional studies would be helpful to elucidate the roles of other CFTR variants (such as the 11T/7T in our patient) in CF pathogenesis and disease phenotype (8).

Compared to classic cases seen in younger patients, adult-onset CF often presents atypically with milder symptoms due to a lower burden of key mutations (9). Using an extensive etiological work-up in 188 adult bronchiectasis patients, CF was identified in 5% of the patients, often with a lag of >15 years between the bronchiectasis and CF diagnoses (10). Other clinical and laboratory characteristics of adult-onset CF cases with late diagnosis include higher frequency of less severe genotypes, lower sweat chloride levels, and reduced incidence of pancreatic insufficiency and Pseudomonas aeruginosa infections (9,11). Importantly, according to data from an Italian registry, about 20% of new CF cases are diagnosed during adult years all the way to the eighth decade of life (12).

Conclusion

Our case highlights the importance of a low threshold for CF evaluation in older adults with unexplained bronchiectasis, frequent pulmonary infections, and the absence of risk factors for lung disease. This case demonstrates key pillars of geriatric care: close multidisciplinary collaboration (multi-complexity), treatment of physical debility via aggressive rehabilitation and arranging supportive services/equipment (mobility), identifying and simplifying complex treatment regimens (medications), and early advanced care planning to align patient values with treatment goals (matters most). Timely recognition of clinical decline and the need for homebound care and hospice are indispensable to help avoid invasive procedures and treatments.

Ethics

Informed Consent: Informed consent for the publication of this case report and any associated clinical details was obtained from the patient's next of kin (her husband, the primary caregiver) and documented in the Electronic Medical Record.

Footnotes

Authorship Contributions

Surgical and Medical Practices: J.L., M.G., Concept: J.L., M.G., Design: J.L., M.G., Data Collection or Processing: J.L., M.G., Analysis or Interpretation: J.L., M.G., Literature Search: J.L., M.G., Writing: J.L., M.G.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.

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