From Missed to Mobilized

Driving Change in Late Cystic Fibrosis Diagnosis

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INTRODUCTION

The CF Project Group for Late-CF Diagnosis is a patient-driven initiative established with the support of Halite Solutions Group (HSG), a nonprofit organization dedicated to improving the quality of life for individuals effected by cystic fibrosis (CF). Operating within HSG's strategic framework, Pathways to Impact prioritizes advancing collaboration, education, and advocacy to meet the specific needs of individuals with CF. This work has received recognition and endorsement from the Aging with CF Special Interest Group.

agingwithCF
a program of halite solutions group

WHO WE ARE

CF Project Group of Late-Diagnosed Individuals
As adults living with cystic fibrosis, many of us received our diagnoses later in life following extended periods of unaddressed symptoms — such as chronic cough, chronic sinus infections, digestive complications, pancreatitis, infertility — and frequent misdiagnoses, including asthma, bronchitis, irritable bowel syndrome, celiac disease, depression, or attributing issues to stress. These symptoms were often addressed individually, without identification of the underlying cause.

Informed by both the care received and the care missing, our experiences offer powerful insights that can inform future care strategies. It is imperative that our backgrounds are recognized and that enhanced support for individuals diagnosed with CF later in life is prioritized across clinical practice, research, and policy advancement.

THE PROBLEM

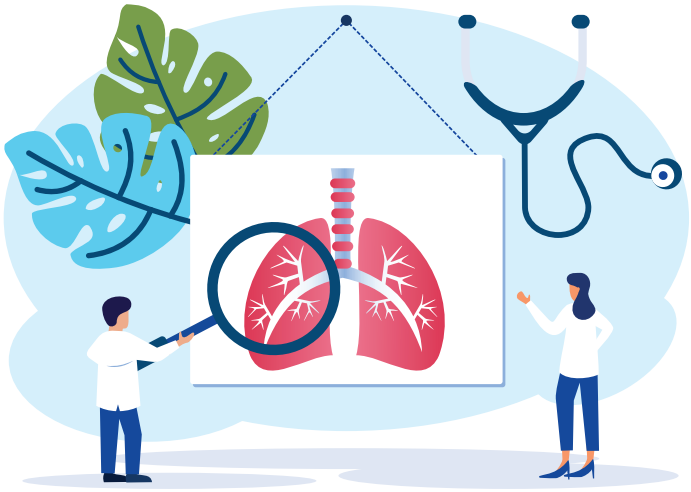
- **Delayed or missed diagnoses:** Adults experience unnecessary health deterioration before a correct diagnosis.
- **Fragmented care:** Multiple specialists treat isolated symptoms without coordination.
- **Psychosocial impact:** Anxiety, depression, and uncertainty are common.
- **Limited awareness:** Many healthcare providers are unaware that CF can present in adulthood.

The delay of diagnosis and subsequent treatment of CF can manifest as debilitating and devastating outcomes for adults experiencing symptoms without proper medical care. Advanced therapies such as CFTR modulators can improve pulmonary function, reduce infection risk and even improve the quality of life for individuals with CF (Barry et al., 2023).

Fragmented care and broken lines of communication impact adult-diagnosed CF individuals with pulmonary symptoms the most. According to the Canadian CF Registry, older adults displaying pulmonary symptoms at the time of diagnosis experience a more rapid decline in lung function compared to other groups of individuals with CF (Desai et al., 2021).

Many individuals diagnosed with CF in adulthood experience a plethora of emotions around the time of diagnosis and even during the aftermath. Feelings of confusion, anxiety, depression, anger and more stem from poorly delivered bad news by physicians, limited education regarding CF at the time of diagnosis, and outdated knowledge transmitted by non-pediatric physicians. The psychosocial turmoil surrounding this pivotal moment needs to be better managed to improve our trust of providers and to enhance adherence to vital treatment (Widerman, 2002).

Many healthcare providers are unaware that CF can present in adulthood. This is revealed by the numerous accounts of individuals diagnosed in later decades of life which is exemplified by the story of a 60-year-old man covered in the Annals of Allergy, Asthma and Immunology in 2000. This case study highlights how individuals with asthma associated with allergic bronchopulmonary aspergillosis and details from past medical history of CF should prompt providers to have a high index of suspicion for CF (Najib et al., 2000).



OUR STORY

Over many years, our experiences have been marked by missed opportunities, ongoing misdiagnoses, and challenges in obtaining recognition as a distinct population with separate needs from the pediatric CF population. While demonstrating resilience, these experiences also underscore systematic shortcomings that warrant attention through increased awareness, comprehensive education, and policy reform.

Our diagnoses were delayed not because symptoms were absent, but because cystic fibrosis was not initially considered. Numerous individuals did not fit conventional diagnostic profiles and were inadvertently disregarded because of outdated perceptions and surface level exposure throughout medical education about CF.

Enduring years of unexplained illness has significantly influenced our perspective and reinforced our resolve to drive systemic improvement. Drawing on these experiences, we are dedicated to advancing the healthcare system to ensure that future adults receive early recognition, accurate diagnoses, and comprehensive care. Our objective is to prevent others from facing the delays, misdiagnoses, and service gaps that we have encountered.

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WHY IS THIS HAPPENING?

We didn't fit the textbook. We weren't newborns. We didn't have failure to thrive (FTT) in the first decade of life like the stereotypical individual with CF. We were adults with physical attributes, mature ages, and broad, atypical medical histories that didn't fit the picture most physicians associate with cystic fibrosis.

A discordance between individuals with CF and the timely, accurate diagnosis has proven not only to have lasting implications on patient care but also to be real and life-altering. Adult physicians are often unfamiliar with the extensive breadth of CF and the various clinical presentations. Specialists are unsure how to support adults with late or atypical diagnoses. The line of communication between doctors is broken, leaving us to act as our own care coordinators, advocates, and educators. The CF system wasn't designed for older adults, and adult medicine wasn't developed to detect conditions primarily associated with the pediatric population.

What comes next needs to look vastly different. We need diagnostic criteria that reflect adult clinical presentations. We need awareness that CF doesn't only happen in childhood, but spans decades beyond the perceived life expectancy of those with CF. Cross-specialty collaboration and interdisciplinary care are essential since adults with CF present with multi-system issues. CF care models must account for aging and long-term survivorship. We also need to include more diverse voices in all aspects of CF, as stories like ours are still too often overlooked and pigeonholed into a monochromatic representation.

To prevent these missed diagnoses for others, we are calling for meaningful changes across healthcare, research, and policy.

What We Are Asking For

Top Five Calls to Action

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1 Revise CF Diagnostic Guidelines for Adults

Expand diagnostic algorithms to include adults with persistent, unexplained symptoms such as sinus disease, pancreatitis, infertility, or chronic upper respiratory infections. Emphasize sweat tests as an initial test workup vs a last resort measure in order to avoid misallocating resources as well as causing excessive emotional strain.

2 Train the Adult Healthcare Workforce

Educate primary care providers, specialists, nurses, and allied health professionals on adult CF presentations. Implement mandatory CME programs and training modules.

3 Establish Structured Support for Adults at Risk of Being Missed

Provide health navigators, peer mentoring, and immediate onboarding at the point of diagnosis to ensure adults receive coordinated care by creating a streamlined protocol for the diagnosing physician.

4 Advance Research and Data Collection

Fund studies focused on diagnostic delays, quality of life, and outcomes for adults with postponed diagnoses. Develop a distinct late-diagnosis registry. Ensure clinical trial inclusion in order to further advancement in CF care in addition to adding meaningful value to years lived post-diagnosis.

5 Elevate Voices of Adults with Delayed Diagnoses

Ensure adults diagnosed later in life are represented on CF boards, research committees, advocacy groups, and policy discussions to provide insightful recommendations and perspectives.

THE POWER OF YOUR ACTIONS

Implementing these recommendations will:

- Reduce delays in diagnosis and prevent avoidable health decline.
- Improve access to comprehensive, coordinated care.
- Promote equity and inclusion for a population historically overlooked.
- Ensure that research, medical education and policy decisions reflect the full spectrum of CF experiences.
- Empower patients to shape the future of CF care and advocacy.

By taking these steps, we can transform delayed diagnoses into systemic advocacy, ensuring that adults with CF are recognized early and no one is overlooked.

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Late-CF Diagnosis Project Group

Direct Support

Julie Carel

Maryann P. Kaminsky, M.Ed., CCC-SLP

Loke Keihanaikukauakahihulihe'ekahaunaele-Keanaaina

Mary Suthers

Denise Thayne, M.Ed.

Chris Wayland

Creative, Editorial and Technical Team

Rebecca Preslar,
Graphic Design & Marketing Specialist

Kristina Robinson,
President & Founder, Halite Solutions Group

Bre'anca Sanders, MMS, TGen Helios Scholar 2020,
MD Candidate, Class of 2027, The University of Arizona College of Medicine-Phoenix

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