

Preface

Cystic fibrosis (CF) is a common genetic disease that arises from mutations in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene. More than a thousand mutations in the gene encoding *CFTR* have been reported since it was first identified in 1989. It has been more than a decade since the publication of the last issue of *Clinics in Chest Medicine* dedicated to CF. In that time the understanding of CF disease pathogenesis has evolved, changing the landscape of CF care. Additionally, as the population of patients who have CF ages, clinicians have gained experience with the unique challenges of caring for adults who have CF. The goal of this issue of *Clinics in Chest Medicine* is to provide a “snapshot” of CF in 2007, highlighting what has been learned about disease pathogenesis and new therapies.

Major improvements in outcomes for patients who have CF have been made during the last several decades and result at least in part from improvements in the understanding of disease pathogenesis. Dr. Strausbaugh and Dr. Davis give an overview of CF and review key epidemiologic and pathobiologic factors discovered thus far. This information is followed up by a more in depth discussion by Dr. Morrissey on the pathogenesis of bronchiectasis.

Part of improving outcomes in CF requires early diagnosis and treatment. Newborn screening is an important first step in early diagnosis and has shown benefits in areas of nutrition, cognitive function, pulmonary function, and survival. The current approach to newborn screening and the experience of Wisconsin in this process is the topic of Dr. Rock’s article. Once children who have CF are diagnosed, preservation of lung function is one of the most important goals and is closely associated with airway microbiology. Dr. Lahiri discusses the consequences of early *Pseudomonas*

aeruginosa colonization and the current approach to management including preliminary experience from an ongoing multicenter clinical trial.

Another important consideration in preservation of lung function in CF is nutritional support. Poor nutrition has long been associated with poor pulmonary and survival outcomes in CF, and aggressive nutritional support should be a fundamental component of any treatment regimen. Dr. Milla reviews the epidemiology and current literature that support a link between lung function and nutrition and discusses approaches to treatment.

In addition to aggressive nutritional support, treatment of airway infection and its associated inflammation is essential to preserving lung function in patients who have CF. Dr. Konstan and Dr. Chmiel provide an overview of the inflammatory response in the CF airway and the current understanding of anti-inflammatory therapies in CF. Dr. McArdle and Dr. Talwalkar then review the current data available for long-term macrolide therapy in CF and discuss the clinical benefits and potential host- and pathogen-related explanations for the positive therapeutic effect.

Although the treatment of airway infection and inflammation is the backbone of pulmonary-related CF care, other novel approaches are currently under investigation. Dr. Weiss, Dr. Sueblinvong, and Dr. Suratt review recent developments in gene and stem cell therapies including a discussion of the feasibility and limitations of these approaches.

Given the poor clinical outcomes associated with airway colonization with resistant organisms, infection-control practices to prevent patient-to-patient transmission of these bacteria are becoming increasingly important. Dr. Zuckerman and Dr. Seder outline some of the major historical events that signaled the need to understand better

the mechanisms of infection in CF and discuss general principles of infection control. They then review the current literature on infection control practice, highlighting areas in which future investigation is needed.

As the understanding of CF lung disease pathogenesis has improved, so has the ability to image the chest. Dr. Robinson reviews current chest imaging modalities and discusses CT as a clinical and research tool and its potential to impact the management of CF lung disease.

A consequence of improved outcomes in CF is the aging population of patients who had CF. An important component of CF care now includes transition from a pediatric to an adult care program. Dr. Parker discusses the varied approaches to successful transition and offers advice for developing skills to help adult CF patients attain their best quality of life. Best quality of life for some adults who have CF includes starting a family. Dr. Sueblinvong and Dr. Whittaker review infertility in men and women who have CF and discuss key issues surrounding pregnancy. Once respiratory failure develops, quality of life may be improved by lung transplantation. Dr. Goldberg and Dr. Deykin review current issues in lung transplantation and special considerations for patients who have CF.

Although lung transplantation offers a treatment option and potentially meaningful, longer survival than the patient with CF's own lungs can provide, the best approach is a focus on delaying the need for lung transplant for as long as possible. Early diagnosis, aggressive nutritional support, and appropriate treatment of airway infection and inflammation are all important in improving outcomes. Despite the wide availability of tested therapies and CF centers that provide

specialized multidisciplinary care by highly trained care givers, there is wide variability in outcomes across the United States. Drs. Quinton and O'Connor complete this issue by discussing quality improvement in CF treatment, including public reporting of CF patient registry data and the belief that providing better care using the currently available therapies is a powerful way to improve clinical outcomes.

The CF community can be proud of the great progress that has been made in the last decade, during which median survival has improved from 30 to 38 years of age. However, many opportunities exist to improve on prior successes, and further work is clearly needed. The CF Foundation has dedicated significant resources to promoting drug development and stimulating large-scale quality improvement efforts. With this support and the ongoing dedication of the CF community, helping patients who have CF live healthy and productive lives is more possible than ever before.

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