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Cystic Fibrosis

Edited by
A.K. Webb and F.A. Ratjen



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Cystic Fibrosis

European Respiratory Monograph 35
April 2006

Editor in Chief
K. Larsson

This book is one in a series of European Respiratory Monographs. Each individual issue provides a comprehensive overview of one specific clinical area of respiratory health, communicating information about the most advanced techniques and systems needed to investigate it. It provides factual and useful scientific detail, drawing on specific case studies and looking into the diagnosis and management of individual patients. Previously published titles in this series are listed at the back of this book with details of how they can be purchased.

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CONTENTS

The Guest Editors	vi
Preface	vii
Introduction	viii
1. New insights into the pathophysiology of lung disease in cystic fibrosis patients G. Döring	1
2. Small airways disease in cystic fibrosis K.G. Brownlee	21
3. Atypical cystic fibrosis K.W. Southern	38
4. Disease modifier genes in cystic fibrosis H. Grasemann	50
5. Emerging pathogens in cystic fibrosis N. Høiby, T. Pressler	66
6. Gene therapy for cystic fibrosis: successes and challenges J.C. Davies, E.W.F.W. Alton	79
7. Cystic fibrosis transmembrane conductance regulator pharmacotherapy L.J.V. Galietta	88
8. Cystic fibrosis infection with clonal strains of <i>Pseudomonas aeruginosa</i>: current knowledge and future management A.M. Jones, S.C. Bell	105
9. Noninvasive ventilation cystic fibrosis B. Fauroux	127
10. Difficult issues in the selection of cystic fibrosis patients for lung transplantation P.A. Corris	139
11. Diagnosis and management of cystic fibrosis related low bone mineral density C.S. Haworth, S.L. Elkin	150

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EUROPEAN RESPIRATORY MONOGRAPH

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Preface

He was one of three frail siblings with recurrent diarrhoea, weight loss and a feeble apparition. A boy who, during childhood and adolescence, was not able to compete with other children of his own age and who was unable to take walks due to respiratory symptoms. He suffered from recurrent respiratory infections, haemoptysis and fevers and got frequent abdominal pain and diarrhoea after eating, in particular fatty pork meals. A man who, before the age of 35, was not able to climb the stairs due to dyspnoea and weakness and who spent most of his latter years spent bedridden with characteristic tiredness, dyspnoea and difficulties with sputum expectoration, until his death in 1849 at the age of 39 yrs. He was a composer and a pianist and his name was Frederic Chopin.

It seems highly likely that Chopin suffered and died from cystic fibrosis CF and not tuberculosis as has been claimed. CF is a disease that was unknown during the nineteenth century. Although it was already asserted in German folklore that "a child who tastes salty on being kissed has a poor prognosis" CF became recognised as a disease in the first half of the 1930s, when it was distinguished from celiac disease. In 1945 mucus secretion abnormalities were described and the term mucoviscidosis was suggested. Further development included characterisation of high sweat levels of sodium and chloride and in the 1960s it was established that CF was an autosomal, recessive, inherited disease. Approximately 20 yrs after this the gene responsible for the disease was located on chromosome seven.

When CF was first described the disease was found to be affecting children who died as a result of it within the first few years of their life. However, it became clear that the CF phenotype differs between individuals and this may, in parallel with improved treatment, explain why certain affected individuals appear to have a more beneficial prognosis with prolonged survival. The knowledge regarding CF has increased tremendously during the previous decades. It is now clear that the disease is caused by a mutation in the gene coding for CF transmembrane conductance regulator, an ion channel responsible for chlorine transport in epithelial cells. The treatment has improved substantially and life-time expectancy has increased from approximately 6 months to 30 yrs. Treatment of airway infections and obstructions, nutritional repletion, anti-inflammatory therapy and lung transplantation have contributed to improve survival outcomes with the possibility of gene therapy soon becoming a probable option.

In the present issue of the European Respiratory Monograph current knowledge regarding CF has been acquired by authors who are true specialists in the field. Most aspects of CF have been covered and this Monograph will be an inestimable source of information for pulmonary physicians and scientists within the field.

K. Larsson
Editor in Chief

INTRODUCTION

Cystic Fibrosis (CF) is a lethal disease affecting children, adolescents and mature adults. However, over the past three decades, as a direct consequence of the application of multidisciplinary care delivered by CF centres worldwide, the disease has changed from one of childhood killer into prolonged adult survival.

The basic defect in CF affects all ductal systems of the body and is truly a disease with multiple systemic manifestations. As patients with CF have grown older many unexpected complications have emerged from Pandora's box to further complicate the lives of CF patients and test the management skills of clinicians. Scientific research into this disease has flourished as demonstrated by the increasing number of sessions devoted to this area at respiratory meetings in Europe and North America.

It is, therefore, timely that the *European Respiratory Journal* is publishing a *Monograph* devoted exclusively to CF. The editors have recruited recognised leaders to write about cutting edge topics of interest both to clinicians and scientists in this field. We are grateful to the authors who have produced superb reviews of their various areas of expertise. We invite all those either with an interest or who want to know more about this fascinating disease to read these reviews and use them as a source of reference and a continuing learning curve.