Cystic Fibrosis

FOURTH EDITION

A Guide for Patient and Family

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To all those patients and families who have so enriched our lives,
and have taught us so well (in the words of Si Kahn),
"it's not just what you're given,
but what you do with what you've got."

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PREFACE

I wrote this book (with a lot of help) for everyone with an interest in cystic fibrosis, whether they be patients, the friends and family of patients, or health professionals who work with patients and their families. It is designed to be of particular use to parents in the initial months that follow their child's diagnosis of cystic fibrosis. It is also written with the intention that it serve as a "refresher" course for people to review areas of treatment and physiology that they may have forgotten. The relatives and friends of a patient may also benefit from this introduction to cystic fibrosis.

An important group for whom this book is written is teenagers who were diagnosed in infancy. While teenagers grow up knowing a lot about cystic fibrosis, they seldom receive the in-depth explanation that their parents received immediately upon diagnosis. I hope that teenagers will use this book to learn more about cystic fibrosis. A final goal of this volume is to provide a foundation for understanding cystic fibrosis that will enable patients and families to understand more fully the advances that are being made so rapidly in this field.

I've tried to stress throughout this book that cystic fibrosis is a serious disease, yet it is one that can be effectively controlled for long periods of time in most patients. It is a life-shortening disease, yet it is also one in which the outlook for patients' length and quality of life has improved dramatically in a relatively short time and continues to do so. It is a disease for which there is currently no cure, yet it is one for which treatment is very effective. It is a disease that creates demands on patients and families for daily treatments; it is also one in which the efforts of patients and families can greatly influence the health and quality of life of the patient. It is a disease that is commonly accepted as inhibiting normal life, yet the reality is that most patients go to school, play sports, and grow up accomplishing all the tasks, and experiencing all the joys and sorrows, of childhood, adolescence, and young adulthood. It is my hope that patients and their families will find this volume to be of help in all these stages of life.

Many patients, families, and health professionals have responded generously to the first three editions of this book and have made suggestions that we've tried to incorporate in this edition to make it more useful. I've been extremely fortunate (and honored) to have been able to convince several of the leaders in the fields of CF research and clinical care to help with this edition. For this edition, it has been my great good fortune to be joined by two of my colleagues, Drs Jonathan E. Spahr and Daniel J. Weiner, with whom I get to work every day. Having their collaboration in producing this fourth edition has made it a true pleasure to work on and, I am convinced, has made the final product that you're holding in your hands even better than it would otherwise have been. It is the hope of all of us that patients, families, and health care workers will find this volume useful.

David M. Orenstein, M.D.

ACKNOWLEDGMENTS

Many wonderful people helped make this book possible. I am indeed fortunate that fate put me in Cleveland from 1969 to 1981 and enabled me to be introduced to cystic fibrosis by five of the most outstanding physicians, teachers, and human beings—LeRoy Matthews, Carl Doershuk, Bob Stern, Tom Boat, and Bob Wood. That they would then accept me as a student, resident, fellow, and finally as a partner is among the most wonderful things that one could imagine. They taught me reams about CF and life; they helped me show the importance of communication and helped me develop my skills in the use of the written word; and they showed me what dedication and compassion are. I am forever grateful to them.

Many patients and their families taught me, too, and have given me so much more than I could ever give them. Quite a few have given generously of their time to make suggestions for the first three editions of this book, and their suggestions have been invaluable.

I am especially grateful to the patients (and colleagues) from across the country and overseas who have been kind enough to let me know that the previous editions have been helpful to them. I hope the same will be true for this edition also.

Colleagues within the Antonio J. and Janet Palumbo Cystic Fibrosis Center at Children's Hospital of Pittsburgh have also continued to make my work rewarding, and even joyful. Louise Bauer, our CF nurse educator for the past 23 years, has taught us more, kept us in line, and made every day fun. Our staff of clinical nurses are amazing in their ability to handle a gazillion phone calls and eight doctors with knowledge, efficiency, understanding, and humor. Drs. Joel Weinberg and Joe Pilewski continue to amaze me with their wisdom and excellence in their care of our adult patients. Liz Hartigan, Sandy Hurban, Adrienne Horn, Judy Fulton, and Caitlin Clark make our CF research projects run without a hitch, but not without fun, for patients, families, and staff. I'm convinced that Beth Lytle, Iris Yann, and Kristen Roberts are the best CF dietitians anywhere. Melisa Kennedy organizes my office life with wonderful efficiency, lollipops, and good cheer; she has perhaps the greatest laugh in all of Pennsylvania. Janet Palumbo's vision and generosity helped make it possible for me to continue to devote my energies—and that of our entire center—to CF; she will be sorely missed.

I've been lucky in my work partners over the years; now I'm delighted that two of the most wonderful CF physicians and colleagues, Drs. Jonathan E. Spahr and Daniel J. Weiner, have not only joined our CF Center in Pittsburgh, but also have joined me in writing this fourth edition. It gives me confidence that if I should stop writing when I hit 100, or even before, this book can continue.

My parents taught me the pleasures of language and humor and showed me their gentle, loving way with people. I was blessed with the best siblings on earth,

who have provided me with the best nieces and nephews in the southern (brother) and northern (sister) hemispheres. Then, unexpectedly, Alex came along and brought me—and continues to bring me—profound happiness. She also contributed some pretty terrific siblings, nephews, and nieces, all in the northern hemisphere. Finally, there aren't adequate words to describe the joy and wonder that are ours each day because of Jacob Atticus An Toan Orenstein.

David M. Orenstein

Ditto what he said. We really do have an amazing group of professionals in Pittsburgh doing their best every day to give the most up-to-date and most compassionate care to children and adults with cystic fibrosis. Thank you David O for giving me this opportunity to work with great folks and highlight what I have learned from them.

I would also like to acknowledge my parents, Robert and Diane Spahr and my wife, Laura Spahr. My father is my model for the perfect physician. My mother is my model for the perfect mother. Laura somehow figured out how to be great at the physician *and* mother thing (the complete package).

My kids (Caroline, Dane, and Elyse) are awesome. Jamie Malone, you are an inspiration to people you've never even met. And finally, Garrick Chow, you acknowledged me in your last book, so here's your payback.

Jonathan E. Spahr

My love of children and pediatrics was fostered by my father (Allan Weiner), and my interest in cystic fibrosis was sparked by my mother (Betty Weiner) who performed sweat testing at Children's Hospital of Michigan for many years. I hope that my work does honor to their memories. My mother also introduced me to Dr. Robert Wilmott, who showed me that pediatrics rather than pediatric surgery would fit best with my talents and personality. I am grateful for his mentorship over many years. I am deeply indebted to those who trained me, including Jules Allen and Howard Panitch, from whom I learned to love physiology. On their shoulders have I stood, and, without them, I would not be the doctor I am today. My patients have taught me the rewards of long-term relationships. My fondness for pediatric surgery led me to the love of my life, Dr. Aviva Katz, and together, our lives are greatly enriched by Gabriel, Samuel, Shoshana, and Channah.

Daniel J. Weiner

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INTRODUCTION

David M. Orenstein, Jonathan E. Spahr,
and Daniel J. Weiner

WHAT IS CYSTIC FIBROSIS?

Cystic fibrosis (CF) is a life-shortening, inherited disorder that affects the way in which salt and water move into and out of the body's cells. The most important effects of this problem are in the lungs and the digestive system (especially the pancreas), where thick mucus blocks the small tubes and ducts. The lung problem can lead to progressive blockage, infection, and lung damage, and even death if there is too much damage, while the pancreatic blockage causes poor digestion and poor absorption of food, leading to poor growth and under-nutrition. The sweat glands are also affected, in that they make a much saltier sweat than normal. Anyone reading this book has probably heard about the sweat test used to diagnose CF. Most parts of the body that make mucus are also affected, including the reproductive tract in men and women with CF.

ARE ANY PARTS OF THE BODY *NOT* AFFECTED BY CF?

The list of body parts affected by CF can seem overwhelmingly long. But CF does *not* affect the brain and nervous system (it does *not* cause mental retardation); it does *not* affect the kidneys; it does *not* directly affect the heart; it does *not* affect the muscles; it does *not* affect the blood; and, except in the lungs, it does *not* interfere with the immune system (the body's ability to fight infection).

WHAT CAUSES CYSTIC FIBROSIS?

Cystic fibrosis is an inherited disorder that is present from birth, although signs and symptoms of it may not show up for weeks, months, or even years after birth. Although it is inherited, the parents of a child with CF do not have CF, and most often there is no history of it in the family. We all have two CF genes that determine whether or not we have CF. Both of these CF genes need to be abnormal for us to have CF, and CF is inherited by receiving one abnormal CF gene from each parent. Each parent usually has only one abnormal CF gene, and thus has no sign of CF at all. Cystic fibrosis is very common among white people, and is the most common inherited profoundly life-shortening disease, affecting 1 in every 2500 live babies born. One in 25 people carry a mutation of the CF gene. CF is not caused by anything the parents did—or did not do—during the pregnancy. The only way

to get CF is to inherit one abnormal CF gene from each parent. You cannot "catch" CF; it is not contagious.

HOW LONG DO PEOPLE WITH CYSTIC FIBROSIS LIVE?

It is impossible to predict how long a single patient will live. It *is* possible to give some overall statistics. Just a few decades ago, nearly all children with CF died before they reached 2 years of age. By 2010, the average survival had improved to over 37 years, with many people surviving into their 50s and beyond. Some children do still die with CF, but this is much less common than in past years. For one recent year, the death rate among CF patients under 1 year was 0.007 (meaning a rate of 7 babies dying out of every 1000), for children aged 6 to 7 years, it was 0.004 (a rate of 4 children dying out of every 1000), for 14- to 15-year-olds, it was 0.015 (15 of 1000), and for 23-year-olds, it was 0.048.

There are several important factors that explain the tremendous improvement and that explain why the outlook continues to improve almost year by year. First, CF is a newly recognized disease. (It is not a *new* disease, as is related in Appendix E, but a newly *recognized* disease.) It was not until 1938 that Dr. Dorothy Andersen wrote the first medical paper describing a number of children who had died with digestive problems and lung problems. She was the first to recognize that this was not just a coincidence, but represented a single disease, which she called "cystic fibrosis of the pancreas," because the children she examined after they died all had *cysts* (fluid-filled sacs) and scar tissue (*fibrosis*) replacing almost all the normal tissue of their pancreas. The name has been shortened to cystic fibrosis, but her description helped to lay the foundation for recognizing the disease, and therefore treating children who had it. Around this time, antibiotics were becoming available, and lung infections could be treated to a degree. In 1964, Drs. Carl Doershuk and LeRoy Matthews and their colleagues from Cleveland reported the results of 5 years of a comprehensive treatment program. These results were very much improved over previous results, and most modern treatment programs use the same basic principles which these pioneers used.

In the last 50 years, many new antibiotics have become available, making treatment more effective. Further, knowledge of CF has spread widely, so that now most pediatricians and family doctors are able to recognize the signs and symptoms of CF and are able to give children treatment while it can still be helpful, that is, before there is too much irreversible lung damage. Many babies are now diagnosed within the first weeks of life, because of widespread *newborn screening* programs. Very importantly, a nationwide network of CF centers has grown up, where CF experts deliver state-of-the-art care.

The point here is that the medical world has had good comprehensive treatment programs for patients with CF for only a little more than 50 years. This means that there are virtually no patients with CF who are 50 years old *and* were started on a treatment program in the first year of life. There are more and more teenagers

and people in their 20s and 30s who were started on treatment programs early in life, before their lungs were in bad shape, and many of these young adults are doing extremely well. Therefore, there is every reason to be very optimistic about the future of a youngster diagnosed and started on treatment today. Certainly, while an average survival to age 37 years reflects a tremendous improvement, it is not something to be satisfied with; but this situation is continually improving. And, for the first time ever, there are now treatments going through clinical trials in patients with CF that are aimed at the very basic cellular defects that underlie all the problems in CF. If these treatments prove their promise, they may well add decades more to patients' lives.

CYSTIC FIBROSIS CARE

Medical care of patients with CF is best carried out at 1 of the 117 CF Centers accredited by the Cystic Fibrosis Foundation, in conjunction with your own pediatrician, family doctor, or internist. Doctors are becoming better informed about CF, but it is important to be in touch with the CF experts who stay up-to-date with the quickly changing field that CF has become. These experts are found in CF Centers, and there are also many specially trained professionals (nutritionists, social workers, nurses, respiratory therapists, physical therapists) at these centers with extensive knowledge and experience taking care of people with CF. The record is fairly clear that CF patients whose care is coordinated by a CF Center live longer than those who do not attend a center. With the health care system changing, it may be more difficult to get a referral to a CF Center, but it is important to insist on it.

It is also important to continue to have care from a general pediatrician, internist, or family doctor, who can be very helpful with the non-CF health issues that arise in everyone's life.

RESEARCH AND THE BASIC DEFECT

When the first edition of this book was published just a little over twenty years ago, the basic defect in CF was not known, and the gene was not yet discovered. Much was understood about the kind of problems people with CF have, how to prevent many of those problems, and how to treat the problems that cannot be prevented. But at a very basic chemical level, no one knew exactly what went wrong within the cells of the body to cause the problems that occurred. What this meant for treatment was that the medications and therapies were all directed at *secondary* problems (problems that are themselves caused by the basic defect) and not at the underlying problem itself. Another way of putting it is that there was no *cure* for CF.

Much has changed in the past few years, and our understanding of what goes wrong within and outside the cells of people with CF has increased tremendously. The gene for CF has been found and cloned (produced in the lab); there is now a

"CF mouse," "CF ferret," and a "CF pig," created through genetic engineering, while previously no non-human animal had CF, and we know infinitely more about the alterations of cell functioning caused by CF (the basic defect is discussed in Chapter 1). There are even some experimental treatments that appear to get around the basic problem with the abnormally functioning cells. However, there is not yet a proven treatment that successfully (and safely) undoes the basic defect, and, therefore, there is still no cure for CF. This may change by the time you read this book.

The situation is similar to that of diabetes. It is known that people get sick with diabetes because they don't have enough insulin to control their blood sugar. These people can lead normal lives by taking daily insulin shots, but they still have diabetes and will have it until scientists discover and eliminate the cause of inadequate insulin production.

When the second edition of this book was published just 15 years ago, there was no national network to coordinate clinical research on new CF treatments. Such a network was not really needed then, because the increased knowledge about the basic defect in CF cells had not yet led to possible treatments. How different the story is now! The Therapeutics Development Network of the CF Foundation was established in 1997, and enables the testing of new treatments that have been developed as a result of the explosion of our knowledge of the basic defect. Tremendous progress has been made in the search for the ways to undo or get around the basic defect in CF. This is a very exciting time in CF research, because nearly every month an important piece of the puzzle is discovered and new experimental treatments come to light and enter into collaborative clinical studies. The prospects for ever better treatment in the upcoming years are very bright.

A WORD TO NEWLY DIAGNOSED PATIENTS AND FAMILIES

If you are reading this book because you (or, more likely, your child) have (has) just been diagnosed with cystic fibrosis, this is a hard time for you. Many people in your situation feel panicked, numb, or "spacey." You may be angry, frightened, disbelieving. For some of you, along with the bad feelings, there may also be a sense of relief at having a diagnosis, particularly if you've known something was wrong but couldn't get your fears taken seriously, or couldn't get your questions answered. Or, as is becoming increasingly common, you may have been "blindsided" by the news with a call from your doctor's office, telling you about the abnormal results of a newborn screening test. This may be a time when you don't want to hear any more information, or it may be a time when you want to learn absolutely everything there is to know about CF. However you are feeling, it may be a little hard to take in a lot of new information. However you are feeling, you can be certain that there have been many, many people who have experienced these same feelings. It may be helpful to talk about how you're feeling with people in the CF

Center, and in some cases with other families who have been through what you're going through now. The people in the Center can help you find such people if you're interested. As you learn more about CF, and get used to the idea that you (your child) have (has) it, and as you see that in most cases people can live quite a normal childhood, adolescence, and beyond, your emotions will become less raw and times will be less hard.

HOW CAN PEOPLE LEARN ABOUT CYSTIC FIBROSIS?

The purpose of this book is to help you learn about all aspects of CF, including how it is inherited, the problems it causes, how it is treated, and current research. Cystic fibrosis centers and the Cystic Fibrosis Foundation can provide information also. Some of you will want to plow through the book cover-to-cover now, while others may not be able to face even the first chapter just yet. But the book will be here when you're ready for it and can certainly be referred back to when a new question comes up, or if you find you've forgotten something. Encyclopedias and many general medical books are *not* a good source of information, since they are likely to be outdated. Newspapers, especially the tabloids we all see in the checkout line in the grocery store, are also not good sources, for they are likely to announce the discovery of a cure that bears little relation to medical truth. Even if you hear something that sounds encouraging on a national TV news show, be sure to check it out with your CF Center or someone who is knowledgeable and up-to-date on research developments.

There have been several instances of incorrect information—even dangerous information—being reported as medical truth on supposedly reputable news shows. In one of these instances, it was announced that CF was caused by a deficiency of *selenium* (a mineral we all need and one that most of us—with or without CF—get plenty of in the diet), and that a cure existed in taking huge doses of selenium. Several babies died as a result of that report, after being given massive overdoses of selenium. Usually, information about CF that appears in the news is not harmful and is even fairly accurate. But it is wise to be cautious about "dramatic breakthroughs" that are announced. Most often, medical progress is made not by dramatic breakthroughs but rather by tiny steps, with one group of scientists building upon the work of previous researchers. Your CF Center and the CF Foundation are informed of all the reputable work in the field worldwide and will be happy to provide you with this information.

The Internet has many sites related to CF, some of them excellent. But anyone who has had any experience "surfing the net" knows that alongside superb sources of information, there can be not-so-reliable (and sometimes even "wacko") sources. This rule holds true for CF-related sites as well. In Appendix F, you can find a number of web sites that should be reliable. As with any other information source, be sure to check any new information you're not sure about (and maybe some that you *are* sure about) with CF Center staff.

ORGANIZATION OF THE BOOK

The goal of this book is to cover all the important topics that concern people with CF and their families. The opening chapter discusses the basic defect in the cells of people with CF, going over some amazing discoveries that have been made just within the past few years. (Our co-author on this chapter, Dr. Ray Frizzell, is responsible for a lot of the exciting research that is unlocking the secrets of the cellular abnormalities in CF.) Next comes a short chapter summarizing how the diagnosis of CF is made. The respiratory system (lungs), how it normally works, the changes brought about by CF, and the treatment of the lung problems are the subject of Chapter 3. Chapter 4, on the digestive and gastrointestinal system, also reviews both normal functioning and that affected by cystic fibrosis. After these sections, there is a brief chapter on the other body systems affected by cystic fibrosis. Then follows a chapter on nutrition.

Chapter 7 discusses hospitalization and other types of elaborate treatments and is followed by a chapter on organ transplantation for CF (mostly about lung transplantation and a bit about liver transplantation), and then a chapter dealing with various aspects of daily life including day care, school, sports, home responsibilities, and travel. Exercise is considered separately in the following chapter. Next is a chapter on the genetics of CF, which describes the manner in which CF is inherited and a lot of the very new information about molecular genetics, how they determine the abnormalities seen in CF, and even prospects for gene therapy. (One of our co-authors on this chapter, Dr. Garry Cutting, is one of the most prominent experts in CF genetics.)

We then switch gears for a chapter that deals with emotional and psychological issues (growing up with CF, effects on the family of a child with CF, etc.). Teenagers get their own chapter—Chapter 13. The special problems of the adult with cystic fibrosis are discussed in Chapter 14, where we're joined by Dr. Joe Pilewski, one of our colleagues here in Pittsburgh, and the co-director of our own adult CF program. The next chapter discusses the difficult issues surrounding dying with CF. For this chapter, we've been joined by Dr. Elisabeth Dellon from the University of North Carolina, who is recognized for her work with dying patients as well as with patients doing well.

Research—past, present, and future—and some speculation about future treatments are the subjects of the next chapter (co-authored by Dr. Christopher Penland, the Director of Research Programs for the CF Foundation). The national Cystic Fibrosis Foundation is discussed by its President and Chief Executive Officer, Dr. Robert Beall, in the final chapter.

The volume includes several appendices: a glossary of technical terms; a listing of commonly used medications, giving brand names and generic names, uses, and side effects; diagrams illustrating the proper techniques for performing chest physical therapy, and discussions of other airway clearance techniques; a short but chubby group of high-calorie recipes; a brief appendix on major historic landmarks in CF; a list of CF Centers in the United States; and a list of CF Centers worldwide.

Finally, the last appendix is a brief bibliography of outstanding readings (mostly technical) on CF and listing of web sites on the Internet dealing directly or indirectly with CF.

A FINAL NOTE ON THE ORGANIZATION AND CONTENT OF THIS BOOK

Each of the chapters starts with a section labeled The Basics, which includes a few of the most important points of that chapter. Some chapters are very long and have much more detail than you'll need or want at any one time. Some of the science presented—particularly in Chapter 1 (*The Basic Defect*) and in parts of Chapter 11 (*Genetics*)—is *very* difficult to understand, and can be daunting, especially the first time 'round. Try not to be intimidated by it, but keep in mind that it's been hard even for most physicians to keep up with the torrid pace of CF research, and it's taken some of us months or years to become comfortable with these concepts.

For each chapter, The Basics may give you an idea of what's there, and you can skim the chapter for what you want to get out of it. The details will be there when you want them.