Living Well with Dystonia
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A Patient Guide

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To my friend, Victor Passy, whose loyal friendship I cherish; To Richard Obosky, Victor and Janie Tsao, Hedy Orden, Suzanne Mellor, Robert Farmer, James Ruetz, Khalique Khan, Greg Jones, and Donald and Darlene Yoshikawa, for whose support I am grateful; to all my patients from whom I’ve learned so much.

—DDT

To Bharti, who accorded me time, patience, and tolerance.

—MSP

To all my dystonia patients who have taught me as much as I have treated them and to Justin and Vi of the NSTA for their unending and tireless dedication helping those with dystonia.

—KF
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Foreword

Dystonia is a life-altering disorder. No question about that. And it has changed the lives of hundreds of thousands of Americans. The earliest symptoms of dystonia begin subtly and may initially be dismissed as the result of fatigue or a minor injury. Understandably, concerns rise as the symptoms persist or get worse. Many people receive the diagnosis after numerous visits to different physicians. They go from doctor to doctor, explaining over and over again what is going on with their bodies. Finally, a diagnosis emerges, and this moment is usually the first time people have ever heard of this disorder: “Dystonia-what?” Now they know what it is called, but what is it?

Living with dystonia often means learning to live in a new body. It may mean learning to manage a great deal of pain—pain that often limits what you can do. It may mean having to give up simple daily activities or changing your career.

And yet, many people who have been diagnosed with dystonia have not allowed it to control their lives. They have discovered new areas of interest and found new activities that they can enjoy with family and friends. They have resolved to learn more about their bodies and to educate themselves on what dystonia is and how it is treated. After the initial shock and concern over the diagnosis, they come to discover that they are stronger and more resilient than they thought. These persons are an inspiration to all of us.

Studies have shown that when people managing a chronic condition educate themselves, they do better. When you take the time to learn all you can—especially from the conversations you have with your doctor—you become an active part of your own treatment plan.

Treating dystonia is an art as well as a science. What works to relieve symptoms for one person with dystonia may not work for another. It may take a couple of tries to discover the most beneficial
treatment plan. You and your doctor must work together to find what works best for you.

We hope you will use this book as a resource for you and your family. It is a tool to help you become better educated about your dystonia and the breadth of available treatments. It provides an overview of what dystonia is, how it is diagnosed and treated, and suggestions on what other things you and your doctor might consider doing to help you with your dystonia. Get informed, stay informed, and get connected. Join a support group or find a forum online. And know you are not alone in this. These steps will help you in your battle with dystonia.

Janet L. Hieshetter
Executive Director
Dystonia Medical Research Foundation
Preface

Twenty years ago, dystonia was not a well known disorder, even to neurologists. But, thanks to the efforts of patient organizations, it is no longer obscure and has even developed into a subspecialty of neurology. It is, however, still vaguely recognized by most physicians who are not neurologists. Many of our patients lived with their disorder for years before receiving a definitive diagnosis. Additionally, there remains a lack of information outside of the professional medical literature for use by affected individuals and their families. It is for this reason, to develop educational and reference materials appropriate for laypersons, that we wrote our first book, The Handbook of Spasmodic Torticollis, in conjunction with the National Spasmodic Torticollis Association. That book, intended for persons afflicted with one particular type of dystonia (spasmodic torticollis, also known as cervical dystonia), sold well. In recent years, the understanding of dystonia and its treatments have markedly expanded. Living Well With Dystonia includes expanded sections on the various forms of the disorder.

This book provides comprehensive information for patients with all kinds of dystonias and the people close to them. Medical terms and concepts are introduced sequentially and then used as building blocks for the later discussion. Although we have provided an extensive glossary, we suggest that you read the chapters in order.

Our intent is to present a clear definition of each dystonia, categorize it appropriately as a movement disorder that is part of the broader category of neurologic diseases, and differentiate it from other conditions with which it may be confused. We discuss different types of dystonia and present the relevant anatomy and physiology of the nervous system. We have drawn on the stories of our patients to build a progressive depiction of the experiences an individual might have as he or she goes through the initial onset of symptoms,
progression of the disorder, finding good medical care, receiving a diagnosis, undergoing treatment, and subsequent outcome. Personal vignettes from the experiences of selected patients are provided to illustrate particular points in the discussion. Subsequent chapters discuss various modes of treatment for dystonia. Chapters 13 and 15 are largely reprinted from our previous book.

Prior to the mid-1980s, there were no specific treatments for dystonia. Nearly all treatment consisted of using oral medications that were primarily intended for other medical conditions. Most of these medications are still in use, and a few new ones have been added. Chapter 10 details and discusses the general principles of medication therapy. During the past decade, a more specific intervention, chemodenervation using botulinum toxin, has become the primary and most effective treatment for some forms of dystonia. More familiar to the public as a cosmetic treatment for wrinkles, there is little awareness of botulinum toxin as a treatment for movement disorders. We have addressed this problem in Chapter 11. Additionally, for those few patients requiring deep brain stimulation surgery, we provide a description of neurosurgical techniques in Chapter 12.

Chapter 15 is a manual of therapeutic rehabilitation exercises. Our patients repeatedly ask us if there are any exercises or rehabilitative techniques they can follow to help alleviate their symptoms. For these reasons, we developed a group of exercises that can be performed by most patients without assistance and using a bare minimum of equipment. We first presented these exercises in a videotape, Physical Therapy and Exercises for Spasmodic Torticollis. In this book, we present these exercises in the same order and format used in the videotape. This book may be used alone to learn the exercises or in conjunction with the videotape.

Since each person’s case of spasmodic torticollis is different, only certain exercises are appropriate for them. We advise you to read through each of the exercises in the chapter. Each exercise contains a description of the particular muscles and type of abnormal neck position that it is intended to treat. You can thus identify which exercises are appropriate for you and discuss the exercises with your physical therapist if you have one. Please obtain permission from your doctor before beginning any of the exercises. The figures in this
book were drawn by Dr. Mayank Pathak and Dr. Hiep Truong, who have strived to accurately represent subtle differences that pertain to different forms of dystonia without making the drawings either too complex or too simple.

Daniel Truong
Mayank Pathak
Karen Frei
In 1986, I had a chance to share a bus ride to a dystonia retreat outside of New York City with a remarkable woman. I was a fellow in my first year in Movement Disorders, a new field of neurology. The motherly woman who spoke with a Texan drawl gently told me the story of a life with blepharospasm. I do not remember now how long the bus ride was. For me it went quickly as she was such a fascinating, inspiring woman. I learned so much about blepharospasm from her. She was an amazing woman with a disarming gentle charm.

After being diagnosed with blepharospasm in 1980, Mattie Lou Koster of Beaumont, Texas, decided to found the Benign Essential Blepharospasm Research Foundation (BEBRF) at the age of 68. In addition from suffering from the symptoms and associated difficulties of the disorder, she had recently lost her husband of 50 years. Koster was subsequently diagnosed with leukemia, breast cancer, and peripheral neuropathy. She later underwent a mastectomy and two hip replacements. Indeed, Koster was an amazingly courageous and resolute woman who overcame many obstacles to fulfill her mission of advancing knowledge about blepharospasm and providing support to people with the disorder. Koster was extremely dedicated to the BEBRF. She knew she was not the only person dealing with blepharospasm and wanted to help others with the disorder. With the knowledge that blepharospasm patients at the time felt isolated and struggled on a daily basis to deal with their symptoms, Koster founded the BEBRF with the intent to help them and to make the disorder more widely known. She was so dedicated to helping people with blepharospasm that, even as the BEBRF expanded, she had calls directed to her personal phone when the office lines were not open so that she could immediately assist anyone who needed help. The organization was originally run from Koster’s home office, where she wrote and mailed newsletters and took calls. For the first six
years, the BEBRF was staffed entirely by volunteers and supported only by small donations from locals and Koster’s own funds.

The BEBRF eventually became widely renowned due in large part to Koster’s efforts. She wrote to a number of medical schools throughout the United States to request more information on blepharospasm and also spoke to any doctors who crossed her path. The first major advance Koster made in spreading knowledge about blepharospasm was when she gave a 10-minute speech to the American Academy of Ophthalmology. The next day, she attended the first of many press conferences that would make an increasingly greater number of doctors aware of blepharospasm. For the same reason Koster also appeared at physicians’ meetings nationwide and subsequently introduced many doctors to using botulinum toxin to treat blepharospasm. She even volunteered to be the first human subject to undergo an experimental procedure developed by Dr. Jonathan Wirtschafter, called chemomyectomy. It involved a series of injections around the eye to paralyze some muscles permanently and theoretically reduce or eliminate blepharospasm symptoms.

Another of Koster’s achievements was to bring together blepharospasm patients from around the world. The newsletters, pamphlets, and other materials that her organization published were influential in making contact with many people with the disorder.

When shipments of botulinum toxin were suddenly suspended and blepharospasm patients were left without any form of treatment, Koster wrote an article in the BEBRF newsletter to encourage her readers to write to the U.S. Food and Drug Administration to resume distribution of the toxin. She managed to organize many people affected by blepharospasm around an important cause and eventually shipments of botulinum toxin resumed.

Another significant occurrence that united blepharospasm patients was when the Wall Street Journal published a front-page article on the disorder in January 1982. Suddenly hundreds of people realized that blepharospasm was the cause of their symptoms and many physicians learned of the disorder for the first time. Koster was also very successful in organizing symposiums about blepharospasm. She always made sure to invite doctors that specialized in either ophthalmology or neurology, since both subjects are relevant to blepharospasm. This provision also ensured the opportunity for
neurologists to interact with ophthalmologists and vice versa, which in turn generated ideas about better treatments for blepharospasm.

Mattie Lou Koster was a forceful and motivated woman who devoted all of her time and energy to the BEBRF and its mission of helping blepharospasm patients and distributing information about the disorder. Her efforts allowed the BEBRF to rise from its humble origins to become an invaluable resource to all people with blepharospasm.

Daniel Truong
Acknowledgments

The authors have not personally suffered with any type of dystonia; we thus acknowledge and give profuse thanks to our patients, whose personal accounts of their illnesses have lent this book a personal touch from a first-hand perspective.

We also acknowledge and thank those of our patients who gave us their personal experiences to use as vignettes, and all of those and their loved ones from whom we have gleaned knowledge over the years. Our special thanks go to Donna Ball, Barbara Barry, Wanda Glaze, Brita Goldsmith, Kathrin Hogan, Dave Jones, Greg Jones, Khalique Khan, Edward Kho, Suzanne Mellor, Ruby Netzley, Lee Ann Orrick, Leslie Rodriguez, Jim Ruetz and Jenine Flood-Ruetz, Dot Sowerby, Darlene and Kristi Yoshikawa for their stories.

We also thank Dr. Hiep Truong for drawing the pictures in Chapter 7.
Introduction

If you are reading this, you or someone close to you has probably been diagnosed with some type of dystonia. Dystonia can take many forms, but all are characterized by involuntary muscle contractions that lead to abnormal postures or involuntary repetitive movements. Dystonias are relatively uncommon disorders, and many physicians—even some neurologists—don’t know much about them. Patients with some dystonias are often misdiagnosed at least once. Patience and perseverance are important in getting an accurate diagnosis and the best care.

Gathering information is the first step in managing any chronic disorder. Finding a specialist in movement disorders or in treating your kind of dystonia, educating yourself about your disorder, and talking to other people with the same diagnosis are all essential to finding the best treatment and achieving the best quality of life.

In this book we provide both the patient’s perspective and the medical perspective to help you begin educating yourself about dystonia. You will learn about factors that may cause dystonia, the signs and symptoms of the condition, how it progresses, and how it is treated. This book will serve as a reference for you to share with your doctor and family.

In our years of treating patients with disabling neurologic conditions, we have found that over several years patients seem to triumph over their initial feelings of despair and adapt to their new state of health and ability. You will see examples of this in the “Patient Perspective” sections of some chapters.

Newly diagnosed patients have the most difficulty coping with dystonia. It used to be that they got a ‘cold’ and, when the condition went away, they would get back to being themselves. Now they are faced with something permanent. It is not easy to accept this new reality. The following story is shared by a patient who now takes her condition in stride.
Patient Perspective

At 60 years of age, I now have a very different perspective on my illness than I did when I was 36, when my symptoms first started. Back then, I didn’t know what dystonia was. If asked, I would have guessed dystonia to be an obscure, small, Eastern European country. Yeah... that was then. I was so young, so innocent, so... not used to the pain I would have to learn to live with.

A lot has changed since 1984: year-one “BC” (before cervical dystonia). I have adapted. That is what I do. I adapt. I don’t fight it; that makes it worse. I don’t curse the fates anymore; they are obviously deaf. I don’t ignore it; that definitely does not work. I just adapt, take it on the chin, go with the flow, hop on the bus.

I will share my list of why’s with you. A “why” is really just a “why the heck me and not someone else?” You may pick the one you think is the correct answer, if you like.

1. My parent hit me in the head one time too many.
2. I had convulsions as a child.
3. My sibling landed me in the hospital with a broken clavicle, broken rib, ruptured spleen, and slight concussion.
4. I’m Irish and an Ashkenazi Jew.
5. I was in the car in the middle of the five-car crash, and I was the one who got whiplash.
6. It’s my uncle’s fault: he had Parkinson and that’s in the same family as cervical dystonia.
7. I was in a very unhappy marriage for a very long time and was very depressed when the symptoms started. This would make it my first husband’s fault; a plausible “why” for many.
8. Rare disorders gravitate to me. I can be pretty interesting on paper, if you are a doctor.
9. I had a near-death experience.
10. It is written.

Actually, my symptoms may have started as early as my childhood. I had a twitch in my right eyelid. Everyone said it was because I was nervous. That twitch has come and gone throughout my entire life. Maybe there is something to that?
“BC” I lived a pretty healthy, normal life. I mean that I was not a street urchin or a hermit. I had not done drugs, didn’t go to Woodstock, and was probably considered boring. There came a time though, when, just like in a movie, when all the planets align and everything comes crashing down, I found that very thing happening in my very own life. I had always been the strong, dependable, capable one and I was changing into an anxious, depressed, squirming, self-conscious agoraphobic. It was as if I had been possessed. I no longer had control over my body, my feelings, and I felt very lost, ugly, and alone.

I saw doctors. I saw doctor after doctor. It’s disturbing to be at the mercy of someone who is just as helpless as you are when it comes to doing anything at all to change what you are going through. I was told that I had chronic upper back muscle spasms, that I needed to take more iron, that I was suffering from anxiety as a result of being an adult child of an alcoholic. On and on it went. Nothing changed except that the disease got worse. I was at an all-time low. I couldn’t drive. I was embarrassed to go out and be seen with my tremor and my head pulled almost to my shoulder. I was crying every day, and felt more and more pain as time allowed the muscles in my neck and back to twist, pull, and torture me.

It was 15 years ago when I saw a doctor who said he knew of a doctor who might know what my condition possibly could be. I was jaded: I’d been burnt before. But I was hopeful.

Now, Dr. X is quite the character. Once you get to know him you understand his genius. He became my BFF (best friend forever) as soon as he circled my frightened self while I sat quietly in that office chair listening to him list off the symptoms I had been living with for several years. It was unnerving to hear someone so absolutely right. After that, it was just taking care of business.

Learning I had cervical dystonia was overwhelming at first, yet the feeling of vindication was marvelous. Living with it has been a lifelong challenge. I’ve morphed back and forth through surgery and injections of botulinum toxin. I’ve educated myself. I’ve shared some of my time and talents in hopes of making a small difference to others with the disorder. I have even met and married a man who has dystonia. That part was not a sacrifice. He’s the best thing that has ever happened to me.

Yes, at the age of 60 I do know a lot more than I did at age 36. I have learned how fragile we are, how helplessness can change into empowerment when you work with dedicated and compassionate people. I have no regrets regarding my inability to enjoy some of the “normal” things many others my
age take totally for granted. I’m very grateful that for some reason known only to doctors, they decide to find out “why” and “what can be done” to help people in need. My life would be so different and so much more limited if I had not been fortunate, if I hadn’t crossed paths with my BFF and so many other kind and caring souls. Maybe “It is written.”

Not everyone with dystonia has this patient’s attitude. She moves on and her dystonia becomes a side show, not the main event. Because we admire her attitude, we decided to share her story with you. We hope that the information in this book will help you to achieve your goals and to get the answers you need to live the best life possible with your dystonia.

In the following chapter, we will walk you through a basic understanding of the nervous system, an overview of the different types of dystonia, methods of diagnosis, and treatment. The two subsequent chapters discuss the genetics of dystonia and related diseases or other factors that can cause it. The five chapters after that discuss different dystonia syndromes in detail. Descriptions of different forms of dystonia are intertwined with real life stories. There is a chapter on oral medicines used in dystonia treatment. Botulinum toxin injection, a mainstay in the treatment of dystonia symptoms, is given its own chapter, followed by a chapter on deep brain stimulation, which is becoming the standard of treatment in certain dystonia syndromes. Cervical dystonia, a complicated and relatively common form of dystonia, is allotted a large chapter of its own. A final chapter on coping with the diagnosis and symptoms of dystonia is followed by a section on therapeutic home exercises and appendices listing support groups and other information.