Genetic Counseling for Adult Neurogenetic Disease

Jill S. Goldman
Editor

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A Casebook for Clinicians

Springer

Editor
Jill S. Goldman
Taub Institute
Columbia University Medical Center
New York, NY
USA

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This book would never have been conceived or written without the encouragement of my husband, Lee Goldman. Not only is he a role model for me, but the impetus and support for my professional growth. I, therefore, dedicate this book to Lee and to our children—Jeff, Daniel, and Robyn and their spouses, Abbey and Tobin, who also provided unending and unconditional support while listening to me complain.

Accompanying Video

The video clips that accompany this book feature aspects of genetic counseling for several of the neurogenetic conditions discussed in the book. Please refer to these video clips as appropriate. Dr. Sampson's full neurological examination may be of particular interest for genetic counselors who have not been exposed to neurology.

The video clips include both segments of actual genetic counseling sessions and counseling simulations. The clips intend to show some of the unique aspects of counseling for these diseases. We cover diagnostic genetic testing, presymptomatic testing, return of results, impact of testing on family members, genetic research studies, reproductive counseling, and treatment and management. Additionally, a video clip demonstrates the neurological examination. Because of limited access to patients, we could not film every disease discussed in this book.

Participants in the video:
Jill S. Goldman, M.S., M.Phil., C.G.C.
Jacinda B. Sampson, M.D., Ph.D.
Ashley L. Wilson, M.S., C.G.C.
Megan Truitt Cho, Sc.M., C.G.C.
Sarah E. Teed
Kara Ansett
Gracious patients

Preface

Adult neurogenetic disease is a rapidly expanding specialty. Few genetic counseling training programs are able to provide the clinical experience necessary to understand the intricacies of counseling for these diverse diseases. Likewise few neurology training programs provide experience in genetic counseling. The goal of this book is to introduce genetic counselors, neurologists, and other health professionals providing counseling for patients with neurogenetic disease to some of the issues that transpire during counseling sessions for these diseases. We have chosen to focus on adult conditions because genetic counseling students have much more exposure to pediatric disease and because these conditions raise very different problems. Although we provide an overview of each condition's symptoms, diagnosis, management, and genetics, our focus is on the counseling. In part, this is because the field is changing so rapidly that genetic information needs continual updating (and therefore, sources such as GeneReviews and PubMed should be consulted regularly when seeing these patients) and, in part, because there is a lack of resources about genetic counseling for these diseases. Chapter 25 provides readers with descriptions of the neurological examination and neuropsychological evaluation. Please refer to them as you make your way through the chapters.

The book is divided by subspecialty areas. We do not attempt to cover every disease in each area, but rather include those diseases that are more common and have their own particular counseling complexities. Nevertheless, although these diseases have unique issues, many aspects of the counseling discussions can apply to all adult neurogenetic disease as well as to other non-neurological genetic diseases. The accompanying video clips are intended to highlight some of the unique features of these diseases, including symptoms and counseling issues.

The case histories have been altered to protect confidentiality. However, they represent experiences that the authors have found to be compelling and challenging. We hope that they will evoke discussion and provide the reader with an insight into adult neurogenetics.

Even as this book was being written, available genetic testing technologies have changed and new genes have been discovered. The genes discussed in this book, thus, represent the more common genetic etiologies known through 2013. Most

chapters concentrate on testing for specific genes, yet as large next-generation sequencing panels (NGS) and whole exome testing (WES) become less expensive, testing methodologies may shift. Keep in mind, however, that bigger is not necessarily better. Single gene testing or small disease-specific panels may be more appropriate with a definitive diagnosis or narrow differential diagnosis. Multiple variants of unknown significance or incidental findings are common with NGS and WES, thus confounding rather than clarifying diagnosis. Ordering physicians and genetic counselors need to understand which genes are meaningful to explore and need to prepare their patients for possible findings. We hope that this book can contribute to that process.

New York, USA Jill S. Goldman

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This book and accompanying video clips would not exist without the remarkable efforts of these coauthors and video contributors. Special thanks go to Dr. Jacinda Sampson for her many contributions to the written document and to the videos. She was truly a savior! For their generous contributions to producing the video clips, special thanks go to my talented videographer, Devin Pinckard, and to the NYC Chapter of the Alzheimer's Association, the Association for Frontotemporal Degeneration, the Parkinson's Disease Foundation, the Children's Tumor Foundation, and to Daniel Goldman. And finally, I offer great appreciation to those patients and families who consented to be filmed.

Contents

List of Contributors

Amanda Bergner Department of Neurology and the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, Baltimore, MD, USA

Matthew Bower Department of Genetics and Metabolism, University of Minnesota Medical Center, Fairview, Minneapolis, MN, USA

Elise Caccappolo Department of Neurology, Columbia University Medical Center, New York, NY, USA

Joline Dalton Paul and Sheila Wellstone Muscular Dystrophy Center, University of Minnesota, Minneapolis, MN, USA

Jamie C. Fong Department of Neurology, Memory and Aging Center, University of California, San Francisco, San Francisco, CA, USA

Jill S. Goldman Taub Institute, Columbia University Medical Center, New York, NY, USA

Alison LaPean Kirschner Department of Neurology, Center for Patient Care and Outcomes Research (PCOR), Medical College of Wisconsin, Milwaukee, WI, USA

Trisha J Multhaupt-Buell Department of Neurology, Breakefield Group, Massachusetts General Hospital, Charlestown, MA, USA

Jacinda B. Sampson Department of Neurology, Columbia University Medical Center, New York, NY, USA

Alice B. Schindler Neurogenetics Branch, NINDS, NIH, Bethesda, MD, USA

Carly E. Siskind Stanford Hospital and Clinics Neuroscience Center, Stanford University, Stanford, CA, USA

Paul Tuite Department of Neurology, University of Minnesota Medical Center, Fairview, Minneapolis, MN, USA

Jeff Waugh Department of Child Neurology, Boston Children's Hospital, Boston, MA, USA

Elisabeth McCarty Wood Center for Neurodegenerative Disease Research, University of Pennsylvania, Philadelphia, PA, USA

Heather Workman Department of Medical Genetics, Dayton Children's Hospital, Dayton, OH, USA

Additional Video Contributors

Ashley Wilson
Megan Truit
Sarah Teed