Michael Danciger was a pioneer in the identification of modifier genes of complex genetic disorders that lead to blindness. The impact of his research and collaborations continues to be felt and expanded. Equally important was his delightful personal quality of leaving an indelible mark on everyone he met and with whom he worked. Michael was a long-time supporter of the RD Symposia. He is missed, and we are honored to dedicate this proceedings volume to him.
Robert “Bob” Barlow was an extremely energetic man who contributed to many areas of research in vision and neurobiological science and held many responsible positions that influenced numerous investigators. He was a long-time supporter of the RD Symposia, as was his wife, Pat, pictured here together at the RD2004 meeting in Australia. He is missed, and we are honored to dedicate this proceedings volume to him.
Preface

The International Symposia on Retinal Degeneration have been held in conjunction with the biennial International Congress of Eye Research (ICER) since 1984. These RD Symposia have allowed basic and clinician scientists from around the world to convene and present their new research findings. They have been organized to allow sufficient time for discussions and one-on-one interactions in a relaxed atmosphere, where international friendships and collaborations can be fostered.

The XIV International Symposium on Retinal Degeneration (also known as RD2010) was held from July 13–17, 2010 at the Fairmont Tremblant Hotel in the resort village of Mont-Tremblant, Quebec, Canada. The meeting brought together 232 basic and clinician scientists, retinal specialists in ophthalmology, and trainees in the field from all parts of the world. In the course of the meeting, 38 platform and 134 poster presentations were given, and a majority of these are presented in this proceedings volume. New discoveries and state-of-the art findings from most research areas in the field of retinal degenerations were presented. This was the largest of all of the RD Symposia, with the greatest number of attendees and presentations.

The RD2010 meeting was highlighted by three special plenary lectures. The first was given by Elise Héon, MD, of the University of Toronto Hospital for Sick Children, Toronto, Ontario, Canada. Dr. Heon discussed “What Bardet-Biedl Syndrome teaches us about ciliopathies.” The second was given by Gregory Hageman, PhD, of the John Moran Eye Center, University of Utah, Salt Lake City, UT. Dr. Hageman described a “New era in the understanding of age-related macular degeneration.” The third plenary lecture was given by Jayakrishna Ambati, MD, of the University of Kentucky, Lexington, KY. Dr. Ambati discussed “Age-related macular degeneration and the other double helix.”

We thank the outstanding management and staff of the beautiful Fairmont Tremblant Hotel for all of their assistance in making this an exceptionally smooth-running conference and a truly memorable experience for all of the attendees. These included, in particular, Isabelle Gilbert, Émilie Normandeau, and Patrick Skelly. We also thank Jonathan Marier of AVW-TELAV for providing audio/visual equipment and services that resulted in a flawless flow of platform presentations. Lastly, we thank Steven LeFort and, particularly, Marie-Chantal Thibault of JPdL Tremblant.
for their planning and implementing transportation of most of the attendees to and from Montreal and the meeting venue in Mont-Tremblant, as well as all aspects of the end-of-meeting Gala at the beautiful “Summit” overlooking Mont-Tremblant and the majestic Laurentian mountains, with a truly Canadian meal, music, and festivities.

The Symposium received international financial support from a number of organizations. We are particularly pleased to thank The Foundation Fighting Blindness, Columbia, Maryland, for its continuing support of this and all previous biennial Symposia, without which we could not have held these important meetings. In addition, for the fifth time, the National Eye Institute of the National Institutes of Health contributed in a major way to the meeting. In the past, funds from these two organizations allowed us to provide 25–35 Travel Awards to young investigators and trainees working in the field of retinal degenerations. However, the response to the Travel Awards program was extraordinary, with 94 applicants, many more than in the past. For this reason, we sought additional support for the Travel Awards program. The Foundation Fighting Blindness-Canada/Institute of Genetics was a major contributor, and for the first time, we turned to industry sponsors and received generous contributions from Novartis Pharma AG, Alcon, Genentech, Inspire Pharmaceuticals, Pfizer, Inc., Genzyme and Bioptigen, Inc. In total, we were able to fund 42 Travel Awards, the largest number ever for these Symposia. Many of the contributing foundations and industry sponsors sent one to several members of their organization to attend the meeting. Their participation and comments in the scientific sessions were instructive to many, offering new perspectives to some of the problems being discussed.

There were two additional “firsts” for the RD Symposia at RD2010. For the first time, there was a commercial exhibitor at the Symposium, Bioptigen, which demonstrated its Spectral Domain Ophthalmic Imaging System (OCT) for small laboratory animals; this was highly instructive for many of the attendees. Second, the world-famous Tremblant International Blues Festival (17th Edition) was held during the RD2010 meeting, immediately adjacent to the Fairmont Tremblant venue. With almost continuous free performances every evening of the Symposium, many groups of attendees enjoyed these together after dinner.

We also acknowledge the diligent and outstanding efforts of Ms. Holly Whiteside, who carried out most of the administrative aspects of the RD2010 Symposium, designed and maintained the meeting website, and participated in the production of this volume. Holly is the Administrative Manager of Dr. Anderson’s laboratory at the University of Oklahoma Health Sciences Center, and she has become the permanent Coordinator for the Retinal Degeneration Symposia. Her dedicated efforts with the Symposia since RD2000 have provided continuity not available previously, and we are deeply indebted to her. Also, Dr. Michael Matthes in Dr. LaVail’s laboratory played a major role in all aspects in the production of this volume, along with the assistance of Ms. Kelly Ahern, in Dr. LaVail’s laboratory.

Recognizing the need to bring younger individuals into the organizational structure of the RD Symposia, at the RD2008 meeting, we invited Drs. John Ash and Christian Grimm to become members of the organizing committee. Thus, instead of
the rotating head organizer working mostly with Holly Whiteside to organize the meeting and prepare the proceedings volume, Dr. Ash assumed equal responsibility with Dr. LaVail for both efforts for RD2010. Dr. Anderson continued in his role as financial administrator for each of the Symposia, working through the Dean McGee Eye Institute, which generously provides the financial responsibility for the meetings and the mechanism for registration of participants. We were pleased to announce at the Gala at RD2010 that our third new member of the organizing committee is Dr. Cathy Bowes Rickman. Dr. Grimm will work closely with Dr. Hollyfield for the RD2012 meeting to be held in Germany.

Finally, we honor the memory of two colleagues who died during the preparation of the RD2010 meeting in 2009 and 2010. Michael Danciger was a great friend to most who attend our RD meetings. Robert Barlow, likewise, was a long-time attendee of the RD Symposia. Both were outstanding scientists and are missed. We dedicate this volume to Michael and Bob.

San Francisco, CA, USA
Oklahoma City, OK, USA
Oklahoma City, OK, USA
Cleveland, OH, USA
Zurich, Switzerland

Matthew M. LaVail
John D. Ash
Robert E. Anderson
Joe G. Hollyfield
Christian Grimm
Matthew M. LaVail, PhD, is Professor of Anatomy and Ophthalmology at the University of California, San Francisco School of Medicine. He received his Ph.D. degree in Anatomy (1969) from the University of Texas Medical Branch in Galveston and was subsequently a postdoctoral fellow at Harvard Medical School. Dr. LaVail was appointed Assistant Professor of Neurology-Neuropathology at Harvard Medical School in 1973. In 1976, he moved to UCSF, where he was appointed Associate Professor of Anatomy. He was appointed to his current position in 1982, and in 1988, he also became Director of the Retinitis Pigmentosa Research Center at UCSF, later named the Kearn Family Center for the Study of Retinal Degeneration. Dr. LaVail has published extensively in the research areas of photoreceptor-retinal pigment epithelial cell interactions, retinal development, circadian events in the retina, genetics of pigmentation and ocular abnormalities, inherited retinal degenerations, light-induced retinal degeneration, and neuroprotective and gene therapy for retinal degenerative diseases. He has identified several naturally occurring murine models of human retinal degenerations and has developed transgenic mouse and rat models of others. He is the author of more than 160 research publications and has edited 14 books on inherited and environmentally induced retinal degenerations. Dr. LaVail has received the Fight for Sight Citation (1976); the Sundial Award from the Retina Foundation (1976); the Friedenwald Award from the Association for Research in Vision and Ophthalmology (ARVO, 1981); two Senior Scientific Investigators Awards from Research to Prevent Blindness (1988 and 1998); a MERIT Award from the National Eye Institute (1989); an Award for Outstanding Contributions to Vision Research from the Alcon Research Institute (1990); the Award of Merit from the Retina Research Foundation (1990); the first John A. Moran Prize for Vision Research from the University of Utah (1997); the first Trustee Award from The Foundation Fighting Blindness (1998); the fourth Llura Liggett Gund Award from the Foundation Fighting Blindness (2007); and he has received the Distinguished Alumnus Award from both his university (University of North Texas) and his graduate school (University of Texas Medical Branch). He has served on the editorial boards of Investigative Ophthalmology and Visual Science and Experimental Eye Research. Dr. LaVail has been an active participant in the
About the Editors

program committee of ARVO and has served as a Trustee (Retinal Cell Biology Section) of ARVO. In 2009, he was appointed an inaugural ARVO Fellow, Gold, of the 12,000-member organization. Dr. LaVail has been a member of the program committee and a Vice President of the International Society for Eye research. He has also served on the Scientific Advisory Board of the Foundation Fighting Blindness since 1973.

John D. Ash, PhD, is Associate Professor of Ophthalmology in the College of Medicine at the University of Oklahoma Health Sciences Center. He is also an adjunct Associate Professor in the Department of Cell Biology, a member of the Oklahoma Center for Neuroscience, and a faculty member of the Dean A. McGee Eye Institute. In 1994, he received his PhD from the Ohio State University Biochemistry Program, and then accepted a postdoctoral fellowship in the Cell Biology Department, Baylor College of Medicine, in Houston, Texas. In 1999, he became Assistant Professor of Ophthalmology at the University of Oklahoma Health Sciences Center, and was promoted to Associate professor in 2006. Dr Ash is also a Visiting Professor of the Dalian Medical University, Dalian China. Dr. Ash has written and published 40 manuscripts including research articles, book chapters and invited reviews related to vision research. He is currently an Executive Editor for Experimental Eye Research, and an Associate Editor of the Journal of Angiogenesis Research. Dr Ash is an active reviewer for Molecular Vision, Investigative Ophthalmology & Visual Science, Experimental Eye Research and Diabetes. In 2009, Dr. Ash received a research award from Hope for Vision, and in 2010 he received a Lew R. Wasserman Merit award from Research to Prevent Blindness, Inc. Dr. Ash has received grants from the National Institutes of Health, the Foundation Fighting Blindness, Research to Prevent Blindness, Inc, Hope for Vision, and the American Diabetes Association. Dr. Ash is currently serving on the Program Committee of the Association for Research in Vision and Ophthalmology. Dr Ash has served on the scientific review panel for Fight For Sight (2005-2008), and is currently serving on the Scientific Advisory Board of the Foundation Fighting Blindness (Columbia, MD) where he chairs the review committee on Novel Medical Therapies Program. He also serves on the scientific review panel for the Macular Degeneration program of the American Health Assistance Foundation (Clarksburg, MD).

Robert E. Anderson, MD, PhD, is George Lynn Cross Research Professor, Dean A. McGee Professor of Ophthalmology, Professor of Cell Biology, and Adjunct Professor of Biochemistry & Molecular Biology and Geriatric Medicine at The University of Oklahoma Health Sciences Center in Oklahoma City, Oklahoma. He is also Director of Research at the Dean A. McGee Eye Institute. He received his Ph.D. in Biochemistry (1968) from Texas A&M University and his M.D. from Baylor College of Medicine in 1975. In 1968, he was a postdoctoral fellow at Oak Ridge Associated Universities. At Baylor, he was appointed Assistant Professor in 1969, Associate Professor in 1976, and Professor in 1981. He joined the faculty of the University of Oklahoma Health Sciences Center in January of 1995. Dr. Anderson served as director of the Oklahoma Center for Neuroscience (1995-1999) and chairman of the Department of Cell Biology (1998-2007). He has received several honorary appointments including Visiting Professor, West China School of Medicine,
Sichuan University, Chengdu, China; Honorary Professorship, Xi’an Jiaotong University, Xi’an, China; and Honorary Professor of Sichuan Medical Science Academy, Sichuan Provincial People’s Hospital, Sichuan, China. Dr. Anderson has received the Sam and Bertha Brochstein Award for Outstanding Achievement in Retina Research from the Retina Research Foundation (1980), and the Dolly Green Award (1982) and two Senior Scientific Investigator Awards (1990 and 1997) from Research to Prevent Blindness, Inc. He received an Award for Outstanding Contributions to Vision Research from the Alcon Research Institute (1985), and the Marjorie Margolin Prize (1994). He has served on the editorial boards of Investigative Ophthalmology and Visual Science, Journal of Neuroscience Research, Neurochemistry International, Current Eye Research, and Experimental Eye Research. Dr. Anderson has published extensively in the areas of lipid metabolism in the retina and biochemistry of retinal degenerations. He has edited 15 books, 14 on retinal degenerations and one on the biochemistry of the eye. Dr. Anderson has received grants from the National Institutes of Health, The Retina Research Foundation, the Foundation Fighting Blindness, and Research to Prevent Blindness, Inc. He has been an active participant in the program committees of the Association for Research in Vision and Ophthalmology (ARVO) and was a trustee representing the Biochemistry and Molecular Biology section. He was named a Gold Fellow by ARVO in 2009 and was awarded the Proctor Medal from ARVO in 2011. He has served on the Vision Research Program Committee and Board of Scientific Counselors of the National Eye Institute and the Board of the Basic and Clinical Science Series of The American Academy of Ophthalmology. Dr. Anderson is a past Councilor, Treasurer, and President of the International Society for Eye Research.

Joe G. Hollyfield, PhD, is Chairman of Ophthalmic Research and the Llura and Gordon Gund Professor of Ophthalmology Research in the Cole Eye Institute at the Cleveland Clinic, Cleveland, Ohio. He received a Ph.D. from the University of Texas at Austin and did postdoctoral work at the Hubrecht Laboratory in Utrecht, The Netherlands. He has held faculty positions at Columbia University College of Physicians and Surgeons in New York City and at Baylor College of Medicine in Houston, Texas. He was Director of the Retinitis Pigmentosa Research Center in The Cullen Eye Institute at Baylor from 1978 until his move to The Cleveland Clinic Foundation in 1995. He is currently Director of the Foundation Fighting Blindness Research Center at the Cleveland Clinic. Dr. Hollyfield has published over 200 papers in the area of cell and developmental biology of the retina and retinal pigment epithelium in both normal and retinal degenerative tissue. He has edited 15 books, 14 on retinal degenerations and one on the structure of the eye. Dr. Hollyfield received the Marjorie W. Margolin Prize (1981, 1994), the Sam and Bertha Brochstein Award (1985) and the Award of Merit in Retina Research (1998) from the Retina Research Foundation; the Olga Keith Weiss Distinguished Scholars’ Award (1981) and two Senior Scientific Investigator Awards (1988, 1994) from Research to Prevent Blindness, Inc.; an award for Outstanding Contributions to Vision Research from the Alcon Research Institute (1987); the Distinguished Alumnus Award (1991) from Hendrix College, Conway, Arkansas; the Endre A. Balazs Prize (1994) from the International Society for Eye Research (ISER), and
the Proctor Medal (2009) from the Association for Research in Vision and Ophthalmology (ARVO). His Proctor lecture was selected for the 2009 Cless “Best of the Best” Award, given by the University of Illinois Eye and Ear Infirmary. He was named an inaugural Gold Fellow by ARVO in 2009. Since 1991 he has been Editor-in-Chief of the journal, Experimental Eye Research, published by Elsevier. Dr. Hollyfield has been active in ARVO, serving on the Program Committee (1976), as Trustee (Retinal Cell Biology, 1989-94), as President (1993-94) and as Immediate Past President (1994-95). He was also President (1988-91) and Secretary (1984-87) of the International Society of Eye Research. He is Chairman of the scientific review panel for the Macular Degeneration program of the American Health Assistance Foundation (Clarksburg, MD), serves on the scientific advisory boards of the Foundation Fighting Blindness (Owings Mills, MD), the Knights Templar Eye Research Foundation (Chicago, IL), the Helen Keller Eye Research Foundation (Birmingham, AL), the South Africa Retinitis Pigmentosa Foundation (Johannesburg, South Africa), and is Co-Chairman of the Medical and Scientific Advisory Board of Retina International (Zurich, Switzerland).

Christian Grimm, PhD, is Professor for Experimental Ophthalmology at the University of Zurich, Switzerland. He received his Ph.D. degree at the Institute for General Microbiology at the University of Berne in 1990. After an initial postdoctoral position in the field of snRNP maturation, Dr. Grimm conducted research at the University of Wisconsin in Madison, WI, where he studied nucleo-cytoplasmic transport of small RNAs. In 1997 Dr. Grimm moved back to Switzerland where he joined the Lab for Retinal Cell Biology in the department of Ophthalmology at the University of Zurich. Dr. Grimm has led the Lab for Retinal Cell Biology since 2006, and was appointed Professor for Experimental Ophthalmology and joined the medical faculty in 2008. Dr. Grimm has published more than 85 original research and review articles, more than 75 in the field of retinal degeneration. His research focuses on molecular mechanisms of photoreceptor cell death and neuroprotection. Dr. Grimm has received the Alfred Vogt Award (2000), the Retinitis Pigmentosa Award of Pro Retina Germany (2003) and the Pfizer Research Award in Neuroscience (2004). He serves on the Editorial Board of Current Eye Research and Experimental Eye Research, and as a Scientific Review Associate for the European Journal of Neuroscience. Dr. Grimm has received research grants from the Swiss National Science Foundation, the European Union, the University of Zurich and several private funding organizations. He serves on the Scientific Advisory Board of the Foundation Fighting Blindness and is Vice Chairman of the Center for Integrative Human Physiology, a priority research program of the University of Zurich.
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