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John D. Ash · Robert E. Anderson
Matthew M. LaVail
Catherine Bowes Rickman
Joe G. Hollyfield · Christian Grimm
Editors

Retinal Degenerative Diseases

Mechanisms and Experimental Therapy

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Dedication

By all standards, the XVII International Symposium on Retinal Degeneration that was held in Kyoto, Japan, was a tremendous success. It is fitting that we dedicate this book of proceedings from the meeting to two pioneering scientists and ophthalmologists from Japan who have led and advocated for this field for many years. This book is dedicated to the careers of Professor Nagahisa Yoshimura, MD, PhD, and Professor Yozo Miyake MD, PhD.



Nagahisa Yoshimura

Yoshimura has extensive experience in clinical retina and retinal research and is known as an advocate for the field. Research interests of Dr. Yoshimura cover a wide range of retinal diseases. Among them, Dr. Yoshimura's group performed an extensive study on polypoidal choroidal vasculopathy (PCV) using new imaging devices in combination with genetic analyses of known age-related macular degeneration (AMD) risk mutations. Dr. Yoshimura's work demonstrated that PCV shares many common risk alleles with AMD, suggesting that PCV is a novel subtype of exudative AMD and not a distinct clinical entity. This important finding overturned a long-standing consensus that PCV was a distinct disease from AMD. Dr. Yoshimura also works to develop patient-derived iPS cells from patients with AMD and other retinal

degenerations to establish induced-RPE cells. This has led to the development of a novel assay system to evaluate iPS-derived RPE cell function.



Yozo Miyake

Miyake has had a long and productive career in ophthalmology. He served as Professor and Chairman of the Department of Ophthalmology, Nagoya University, until 2005. After retiring from Nagoya University, he served as the Director of National Institute of Sensory Organs in Tokyo, before becoming the President of Aichi Medical University in 2010. In addition, he served as the President of the International Society of Clinical Electrophysiology for Vision (ISCEV) from 2000 to 2004.

Dr. Miyake's major contributions to retinal degeneration include the development of ERG for clinical diagnostics. This includes pioneering use of systems for focal macular ERG (Arch Ophthalmol 1986, IOVS 1984, 1989). This allowed precision measurements of oscillatory potentials (Ops) of human macular ERGs for the first time in the history (IOVS 1989). Miyake's focal macular ERG system was in the forefront of the multifocal ERG developed by Sutter in 1994. The development of the focal macular ERG led to Dr. Miyake's most important work, which includes the discovery of "occult macular dystrophy" (OMD) (AJO 198v9, 1996). OMD is not a rare disease, and the focal macular ERG was key for its diagnosis. Dr. Miyake's group succeeded in identifying the gene mutation (RP1L1) causing OMD (Am J Hum Genet, 2010). Because of the substantial contribution of Dr. Miyake and his group, OMD diseases with the RP1L1 gene mutations are now referred to as "Miyake disease" (IOVS 2016). In 2015, the Japanese Ministry of Health, Labor and Welfare for medical care declared Miyake disease as one of the limited diseases for priority financial support.

Another highlight of Miyake's work using ERGs was the establishment of new clinical definitions of congenital stationary night blindness (Arch Ophthalmol 1986). Using a combination patient data and animal experiments, Dr. Miyake concluded that

complete CSNB has the complete defect of ON bipolar cell function, while incomplete CSNB has the incomplete defect of both ON and OFF bipolar cell function. Miyake's hypothesis has been well supported by data from other groups and still stands as the prevailing hypothesis (Nature Genet 1998, 2000).

Preface

The International Symposia on Retinal Degeneration have been held in conjunction with the biennial meeting of the International Society of Eye Research (ISER) 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 substantial time for discussions and one-on-one interactions in a relaxed atmosphere, where international friendships and collaborations can be fostered. The XVII International Symposium on Retinal Degeneration (also known as RD2016) was held from September 19 to 24, 2016, at the Kyoto International Conference Center, in the beautiful and historic city of Kyoto, Japan. The meeting brought together 294 basic and clinician scientists, retinal specialists in ophthalmology, and trainees in the field from all parts of the world.

Abstract submissions to the RD2016 meeting exceeded all expectations, both in quantity and quality. The scientific program covered many aspects of retinal degeneration. The presentations included 42 platform talks and 151 posters. The program consisted of 3 full days of platform talks and 2 evening poster sessions. The RD2016 meeting was highlighted by five special keynote lectures. The first was given by **Robert Marc, PhD**, of the University of Utah, Salt Lake City, Utah. Dr. Marc discussed “Retinal Connectomes and Pathoconnectomes.” The second keynote lecture was given by **José-Alain Sahel, MD, PhD**, Institut de la Vision, Paris, France. Dr. Sahel discussed “Maintaining and Restoring Cone-Mediated Vision in Rod-Cone Degenerations.” The third keynote lecture was given by **Takeshi Iwata, PhD**, National Institute of Sensory Organs Tokyo, Japan. Dr. Iwata discussed “Genetic Factors and Molecular Mechanisms of Early Stage AMD: CNV Mouse and Drusen Primate Models.” The fourth keynote lecture was given by **Yozo Miyake, MD, PhD**, Aichi Medical University, Nagoya, Japan. Dr. Miyake discussed “Establishment of New Concepts in Three Hereditary Retinal Diseases from Japan.” The fifth and final keynote lecture was given by the 2012 Nobel Laureate **Shinya Yamanaka, MD, PhD**, Kyoto University, Kyoto, Japan. Dr. Yamanaka discussed “Recent Progress in iPS Cell Research and Application.” The scientific meeting ended with a “Welcome to RD2018” by local organizer Dr. Peter Humphries, along with the organizers primarily responsible for the meeting, Drs. Cathy Bowes Rickman and Christian Grimm.

We thank the Local Organizing Committee Chair, **Nagahisa Yoshimura**, Kyoto University, and his local organizing committee Drs. **Shuichi Yamamoto**, Chiba University Hospital; **Akira Murakami**, Juntendo University Hospital; **Yoshiro Hotta**, Hamamatsu University School of Medicine; **Mineo Kondo**, Mie University Graduate School of Medicine; **Akihiro Ohira**, Shimane University Faculty of Medicine; and **Masaki Tanito**, Matsue Red Cross Hospital. In addition, we thank the outstanding management and staff of the meeting planning company, JTB Communication Design, for their assistance in making this an exceptionally smooth-running conference and a truly memorable experience for all of the attendees. These included, in particular, **Chika Okuhashi**, **Toshikazu Mogi**, and **Noriko Kataoka**.

The Symposium was able to fund 49 full-ride travel awards given to graduate students, postdocs, and junior faculty. The Travel Awardees were selected on the basis of 9 independent scores of their submitted abstracts, 6 from each of the organizers and 3 from the other members of the Travel Awards Committee for RD2016, Drs. Jacque Duncan, Michelle Pardue, and XianJie Yang. Travel awards were made possible in part by funding from the National Eye Institute of the National Institutes of Health. We are pleased to report this is the eight consecutive symposium in which the NEI has contributed travel awards to support young investigators. Additional awards were provided by generous international financial support from a number of organizations, including The Foundation Fighting Blindness; the BrightFocus Foundation; Pro Retina, Germany; the Fritz Tobler Foundation, Switzerland; and The Harrington Discovery Institute. Many of the contributing foundations sent members of their organizations 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. For the first time, the Symposium held a Funders Lunch Forum where representatives from the Foundation Fighting Blindness, the Harrington Foundation, Fighting Blindness Ireland, BrightFocus Foundation, and the ProRetina Foundation held a panel discussion to discuss funding opportunities from their organizations. This was well received and will likely be repeated in future meetings.

We also acknowledge the diligent and outstanding efforts of Ms. **Holly Whiteside**. Holly is the Administrative Manager of Dr. Anderson's laboratory at the University of Oklahoma Health Sciences Center. Holly has been the RD Symposium Coordinator since 2000, and continues to serve the RD Symposia by providing administrative assistance with financial aspects of the meeting.

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 Oklahoma City, OK, USA
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 Cleveland, OH, USA
 Zurich, Switzerland

John D. Ash
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 Christian Grimm

Travel Awards

We gratefully acknowledge the support from the National Eye Institute, NIH, USA; the Foundation Fighting Blindness, USA; BrightFocus Foundation, USA; Pro Retina, Germany; and the Fritz Tobler Foundation, Switzerland, in their support of 49 travel awards. Eligibility for an award was restricted to graduate students, post-doctoral fellows, instructors, and assistant professors who were actively involved in retinal degeneration research. These awards were based on the quality of the abstract submitted by each applicant. The Travel Awards Committee consisted of nine senior retinal degeneration investigators and was chaired by Catherine Bowes Rickman.

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University of Southern California, Los Angeles, CA, USA

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