CONTENTS

PREFACE ................................................................. vii

ABOUT THE EDITORS ....................................................... ix

PART I MOLECULAR GENETICS AND CANDIDATE GENES

1. GENETIC FACTORS MODIFYING CLINICAL EXPRESSION OF AUTOSOMAL DOMINANT RP ........................................ 3
Stephen P. Daiger, Suma P. Shankar, Alice B. Schindler, Lori S. Sullivan, Sara J. Bowne, Terri M. King, E. Warick Daw, Edwin M. Stone, and John R. Heckenlively

2. DISEASE-ASSOCIATED VARIANTS OF THE ROD-DERIVED CONE VIABILITY FACTOR (RdCVF) IN LEBER CONGENITAL AMAUROSIS. Rod-derived cone viability variants in LCA .......... 9
Sylvain Hanein, Isabelle Perrault, Sylvie Gerber, Hélène Dollfus, Jean-Louis Dufier, Josué Feingold, Arnold Munnich, Shomi Bhattacharya, Josseline Kaplan, José-Alain Sahel, Jean-Michel Rozet, and Thierry Leveillard

3. LEBER CONGENITAL AMAUROSIS: SURVEY OF THE GENETIC HETEROGENEITY, REFINEMENT OF THE CLINICAL DEFINITION AND PHENOTYPE-GENOTYPE CORRELATIONS AS A STRATEGY FOR MOLECULAR DIAGNOSIS. Clinical and molecular survey in LCA ............... 15
Sylvain Hanein, Isabelle Perrault, Sylvie Gerber, Gaëlle Tanguy, Jean-Michel Rozet, and Josseline Kaplan
4. A FIRST LOCUS FOR ISOLATED AUTOSOMAL RECESSIVE OPTIC ATROPHY (ROA1) MAPS TO CHROMOSOME 8q21–q22
Fabienne Barbet, Sylvie Gerber, Sélim Hakiki, Isabelle Perrault, Sylvain Hanein, Dominique Ducroq, Gaëlle Tanguy, Jean-Louis Dufier, Arnold Munnich, Josseline Kaplan, and Jean-Michel Rozet

5. RCC1-LIKE DOMAIN AND ORF15: ESSENTIALS IN RPGR GENE
Zi-Bing Jin, Mutsuko Hayakawa, Akira Murakami, and Nobuhisa Nao-i

6. CHOROIDAL NEOVASCULARIZATION IN PATIENTS WITH ADULT-ONSET FOVEOMACULAR DYSTROPHY CAUSED BY MUTATIONS IN THE RDS/PERIPHERIN GENE
Darius M. Moshfeghi, Zhenglin Yang, Nathan D. Faulkner, Goutam Karan, Sukanya Thirumalaichary, Erik Pearson, Yu Zhao, Thomas Tsai, and Kang Zhang

7. BIOCHEMICAL CHARACTERISATION OF THE C1QTNF5 GENE ASSOCIATED WITH LATE-ONSET RETINAL DEGENERATION. A genetic model of age-related macular degeneration
Xinhua Shu, Brian Tulloch, Alan Lennon, Caroline Hayward, Mary O’Connell, Artur V. Cideciyan, Samuel G. Jacobson, and Alan F. Wright

8. BIEETTI CRYSTALLINE CORNEORETINAL DYSTROPHY ASSOCIATED WITH CYP4V2 GENE MUTATIONS
Makoto Nakamura, Jian Lin, Koji Nishiguchi, Mineo Kondo, Jiro Sugita, and Yozo Miyake

PART II DIAGNOSTIC, CLINICAL, CYTOPATHOLOGICAL AND PHYSIOLOGIC ASPECTS OF RETINAL DEGENERATION

9. FUNDUS APPEARANCE OF CHOROIDEREMIA USING OPTICAL COHERENCE TOMOGRAPHY
Bradley J. Katz, Zhenglin Yang, Marielle Payn, Yin Lin, Yu Zhao, Erik Pearson, Shan Duan, Shin Kamaya, Goutam Karan, and Kang Zhang

10. A2E, A FLUOROPHORE OF RPE LIPOFUSCIN, CAN DESTABILIE MEMBRANE
Janet R. Sparrow, Bolin Cai, Young Pyo Jang, Jilin Zhou, and Koji Nakanishi
11. AMINO-RETINOID COMPOUNDS IN THE HUMAN RETINAL PIGMENT EPITHELIUM .................................................. 69

12. ANNEXINS IN BRUCH’S MEMBRANE AND DRUSEN ....................... 75
Mary E. Rayborn, Hiro Sakaguchi, Karen G. Shadrach, John W. Crabb, and Joe G. Hollyfield

PART III ANIMAL MODELS OF RETINAL DEGENERATION

13. MOLECULAR MECHANISMS OF PHOTORECEPTOR DEGENERATION IN RP CAUSED BY IMPDH1 MUTATIONS ................................ 81
Aileen Aherne, Avril Kennan, Paul F. Kenna, Niamh McNally, G. Jane Farrar, and Pete Humphries

14. BIOCHEMICAL FUNCTION OF THE LCA LINKED PROTEIN, ARYL HYDROCARBON RECEPTOR INTERACTING PROTEIN LIKE–1 (AIPL1). Role of AIPL1 in retina ........................................... 89
Matthew L. Schwartz, James B. Hurley, and Visvanathan Ramamurthy

15. CHARACTERIZATION OF MOUSE MUTANTS WITH ABNORMAL RPE CELLS ........................................ 95
Chun-hong Xia, Haiquan Liu, Meng Wang, Debra Cheung, Alex Park, Yang Yang, Xin Du, Bo Chang, Bruce Beutler, and Xiaohua Gong

16. ROD AND CONE PIGMENT REGENERATION IN RPE65−/− MICE .................................................... 101
Baerbel Rohrer and Rosalie Crouch

17. INITIAL OBSERVATIONS OF KEY FEATURES OF AGE-RELATED MACULAR DEGENERATION IN APOE TARGETED REPLACEMENT MICE .................................................. 109
Goldis Malek, Brian Mace, Peter Saloupis, Donald Schmechel, Dennis Rickman, Patrick Sullivan, and Catherine Bowes Rickman

18. ALTERED RHYTHM OF PHOTORECEPTOR OUTER SEGMENT PHAGOCYTOSIS IN β5 INTEGRIN KNOCKOUT MICE ............. 119
Emeline F. Nandrot and Silvia C. Finnemann
19. LIGHT/DARK TRANSLOCATION OF ALPHATRANSDUCTIN IN MOUSE PHOTORECEPTOR CELLS EXPRESSING G90D MUTANT OPSIN .................................................. 125
Zack A. Nash and Muna I. Naash

20. SLOWED PHOTORESPONSE RECOVERY AND AGE-RELATED DEGENERATION IN CONES LACKING G PROTEIN-COUPLED RECEPTOR KINASE 1 ........................................ 133
Xuemei Zhu, Bruce Brown, Lawrence Rife, and Cheryl M. Craft

21. TRANSGENIC ANIMAL STUDIES OF HUMAN RETINAL DISEASE CAUSED BY MUTATIONS IN PERIPHERIN/RDS .......................... 141
Xi-Qin Ding and Muna I. Naash

22. TRANSGENIC EXPRESSION OF LEUKEMIA INHIBITORY FACTOR INHIBITS BOTH ROD AND CONE GENE EXPRESSION. Gp130 regulates cone gene expression .......................... 147
John D. Ash and Dianca R. Graham

23. A ROLE FOR BHLH TRANSCRIPTION FACTORS IN RETINAL DEGENERATION AND DYSFUNCTION ........................................ 155
Mark E. Pennesi, Debra E. Bramblett, Jang-Hyeon Cho, Ming-Jer Tsai, and Samuel M. Wu

24. CHARACTERISATION OF A MODEL FOR RETINAL NEOVASCULARISATION. VEGF MODEL CHARACTERISATION .................................. 163
Pauline E. van Eeden, Lisa Tee, Wei-Yong Shen, Sherralee Lukehurst, Chooi-May Lai, P. Elizabeth Rakoczy, Lyn D. Beazley, and Sarah A. Dunlop

25. A TWO-ALTERNATIVE FORCED CHOICE METHOD FOR ASSESSING MOUSE VISION ............................................... 169
Yumiko Umino, Bridget Frio, Maryam Abbasi, and Robert Barlow

26. CONDITIONAL GENE KNOCKOUT SYSTEM IN CONE PHOTORECEPTORS ............................... 173
Yun-Zheng Le, John D. Ash, Muayyad R. Al-Ubaidi, Ying Chen, Jian-Xing Ma, and Robert E. Anderson

27. REGULATION OF TIGHT JUNCTION PROTEINS IN CULTURED RETINAL PIGMENT EPITHELIAL CELLS AND IN VEGF OVEREXpressing TRANSGENIC MOUSE RETINAS .......... 179
Reza Ghassemifar, Chooi-May Lai, and P. Elizabeth. Rakoczy
28. PATHOLOGICAL HETEROGENEITY OF VASOPROLIFERATIVE RETINOPATHY IN TRANSGENIC MICE OVEREXPRESSING VASCULAR ENDOTHELIAL GROWTH FACTOR IN PHOTORECEPTORS .......................... 187

29. LASER PHOTOCOAGULATION: OCULAR RESEARCH AND THERAPY IN DIABETIC RETINOPATHY .................. 195
Caroline E. Graham, Nicolette Binz, Wei-Yong Shen, Ian J. Constable, and Elizabeth P. Rakoczy

30. APPLYING TRANSGENIC ZEBRAFISH TECHNOLOGY TO STUDY THE RETINA ....................... 201
Ross F. Collery, Maria L. Cederlund, Vincent A. Smyth, and Breandán N. Kennedy

31. BMI1 LOSS DELAYS PHOTORECEPTOR DEGENERATION IN RD1 MICE. Bmi1 loss and neuroprotection in Rd1 mice ............... 209
Dusan Zencak, sylvain V. Crippa, Meriem Tekaya, Ellen Tanger, Daniel F. Schorderet, Francis L. Munier, Maarten van Lohuizen, and Yvan Arsenijevic

32. TRANSCRIPTIONAL AND POST-TRANSCRIPTIONAL REGULATION OF THE ROD cGMP-PHOSPHODIESTERASE β-SUBUNIT GENE. Recent advances and current concepts ........ 217
Leonid E. Lerner, Natik Piri, and Debora B. Farber

PART IV GENE THERAPY AND NEUROPROTECTION

33. DOWN-REGULATION OF RHODOPSIN GENE EXPRESSION BY AAV-VECTORED SHORT INTERFERING RNA ........ 233
Jacqueline T. Teusner, Alfred S. Lewin, and William W. Hauswirth

34. ASSESSING THE EFFICACY OF GENE THERAPY IN Rpe65⁻/⁻ MICE USING PHOTOENTRAINMENT OF CIRCADIAN RHYTHM .... 239
35. LENTIVIRAL VECTORS CONTAINING A RETINAL PIGMENT EPITHELIUM SPECIFIC PROMOTER FOR LEBER CONGENITAL AMAUROSIS GENE THERAPY. Lentiviral gene therapy for LCA .......................................................... 247
Alexis-Pierre Bemelmans, Corinne Kostic, Dana Hornfeld, Muriel Jaquet, Sylvain V. Crippa, William W. Hauswirth, Janis Lem, Zhongyan Wang, Daniel F. Schorderet, Francis L. Munier, Andreas Wenzel, and Yvan Arsenijevic

36. GENE DELIVERY TO THE RETINA USING LENTIVIRAL VECTORS .......................................................... 255
Kenneth P. Greenberg, Edwin S. Lee, David V. Schaffer, and John G. Flannery

37. POTENTIAL USE OF CELLULAR PROMOTER(S) TO TARGET RPE IN AAV-MEDIATED DELIVERY. Cellular promoters and RPE-targeting .......................................................... 267
Erika N. Sutanto, Dan Zhang, Yvonne K.Y. Lai, Wei-Yong Shen, and P. Elizabeth Rakoczy

38. CYTOKINE-INDUCE RETINAL DEGENERATION: ROLE OF SUPPRESSORS OF CYTOKINE SIGNALING (SOCS) PROTEINS IN PROTECTION OF THE NEURORETINA ............ 275
Charles E. Egwuagu, Cheng-Hong Yu, Rashid M. Mahdi, Maire Mameza, Chikezie Eseonu, Hiroshi Takase, and Samuel Ebong

39. DISEASE MECHANISMS AND GENE THERAPY IN A MOUSE MODEL FOR X-LINKED RETINOSCHISIS .................. 283

40. MOLECULAR MECHANISMS OF NEUROPROTECTION IN THE EYE .......................................................... 291
Colin J. Barnstable and Joyce Tombran-Tink

41. RETINAL DAMAGE CAUSED BY PHOTODYNAMIC THERAPY CAN BE REDUCED USING BDNF .................. 297
Jacque L. Duncan, Daniel M. Paskowitz, George C. Nune, Douglas Yasumura, Haidong Yang, Michael T. Matthes, Marco A. Zarbin, and Matthew M. LaVail
42. CONTROLLING VASCULAR ENDOTHELIAL GROWTH FACTOR: THERAPIES FOR OCULAR DISEASES ASSOCIATED WITH NEOVASCULARIZATION ......................................................... 303
Robert J. Marano and P. Elizabeth Rakoczy

43. INTRAVITREAL INJECTION OF TRIAMCINOLONE ACETONIDE FOR MACULAR EDEMA DUE TO RETINITIS PIGMENTOSA AND OTHER RETINAL DISEASES ........................................... 309

44. CONE SURVIVAL: IDENTIFICATION OF RdCVF ........................................ 315
Olivier Lorentz, José Sahel, Saddek Mohand-Saïd, and Thierry Leveillard

45. NEUROPROTECTION OF PHOTORECEPTORS IN THE RCS RAT AFTER IMPLANTATION OF A SUBRETINAL IMPLANT IN THE SUPERIOR OR INFERIOR RETINA ........................................ 321
Machelle T. Pardue, Michael J. Phillips, Brett Hanzlicek, Hang Yin, Alan Y. Chow, and Sherry L. Ball

46. GLUTAMATE TRANSPORT MODULATION: A POSSIBLE ROLE IN RETINAL NEUROPROTECTION ................................................................. 327
Nigel L. Barnett, Kei Takamoto, and Natalie D. Bull

47. ACTIVATION OF CELL SURVIVAL SIGNALS IN THE GOLDFISH RETINAL GANGLION CELLS AFTER OPTIC NERVE INJURY .......................................................... 333
Yoshiki Koriyama, Keiko Homma, and Satoru Kato

PART V USHER SYNDROME

48. ROLES AND INTERACTIONS OF USHER 1 PROTEINS IN THE OUTER RETINA ................................................................. 341
Concepción Lillo, Junko Kitamoto, and David S. Williams

49. MOLECULAR ANALYSIS OF THE SUPRAMOLECULAR USHER PROTEIN COMPLEX IN THE RETINA. Harmonin as the key protein of the Usher Syndrome ........................................ 349
Jane Reiners and Uwe Wolfrum
PART VI STEM CELLS, TRANSPLANTATION AND RETINAL REPAIR

50. LIMITED NEURAL DIFFERENTIATION OF RETINAL PIGMENT EPITHELIUM
Ryosuke Wakusawa, Toshiaki Abe, Yoko Saigo, and Makoto Tamai

51. RETINAL PIGMENT EPITHELIAL CELLS FROM THERMALLY RESPONSIVE POLYMER-GRAFTED SURFACE REDUCE APOPTOSIS
Toshiaki Abe, Masayoshi Hojo, Yoko Saigo, Masahiko Yamato, Teruo Okano, Ryosuke Wakusawa, and Makoto Tamai

52. RETINAL TRANSPLANTATION. A treatment strategy for retinal degenerative disease
Biju B. Thomas, Robert B. Aramant, SriniVas R. Sadda, and Magdalene J. Seiler

53. MICROARRAY ANALYSIS REVEALS RETINAL STEM CELL CHARACTERISTICS OF THE ADULT HUMAN EYE.
B Brigitte Angénieux, Lydia Michaut, Daniel F. Schorderet, Francis L. Munier, Walter Gehring, and Yvan Arsenijevic

54. USING STEM CELLS TO REPAIR THE DEGENERATE RETINA.
Christine M. Hall, Anthony Kicic, Chooi-May Lai, and P. Elizabeth Rakoczy

55. OPTIC NERVE REGENERATION: MOLECULAR PRE-REQUISITES AND THE ROLE OF TRAINING.
Restoring vision after optic nerve injury
Lyn D. Beazley, Jennifer Rodger, Carolyn E. King, Carole A. Bartlett, Andrew L. Taylor, and Sarah A. Dunlop

56. RETINAL GANGLION CELL REMODELLING IN EXPERIMENTAL GLAUCOMA
James E. Morgan, Amit V. Datta, Jonathan T. Erichsen, Julie Albon, and Michael E. Boulton

PART VII INDUCED RETINAL DEGENERATIONS

57. NEURAL PLASTICITY REVEALED BY LIGHT-INDUCED PHOTORECEPTOR LESIONS
Bryan W. Jones, Robert E. Marc, Carl B. Watt, Dana K. Vaughan, and Daniel T. Organisciak
58. FACTORS UNDERLYING CIRCADIAN DEPENDENT SUSCEPTIBILITY TO LIGHT INDUCED RETINAL DAMAGE .................................................. 411
Ruby Grewal, Daniel T. Organisciak, and Paul Wong

59. SPACE FLIGHT ENVIRONMENT INDUCES DEGENERATION IN THE RETINA OF RAT NEONATES ................................. 417
Joyce Tombran-Tink and Colin J. Barnstable

60. TOXICITY OF HYPEROXIA TO THE RETINA: EVIDENCE FROM THE MOUSE .................................................. 425
Scott Geller, Renata Krowka, Krisztina Valter, and Jonathan Stone

61. TREATMENT WITH CARBONIC ANHYDRASE INHIBITORS DEPRESSES ELECTRORETINOGRAM RESPONSIVENESS IN MICE .................................................. 439
Yves Sauvé, Goutam Karan, Zhenglin Yang, Chunmei Li, Jianbin Hu, and Kang Zhang

62. INJURY-INDUCED RETINAL GANGLION CELL LOSS IN THE NEONATAL RAT RETINA .................................................. 447
Kirsty L. Spalding, Qi Cui, Arunasalam M. Dharmarajan, and Alan R. Harvey

PART VIII BASIC SCIENCE UNDERLYING RETINAL DEGENERATION

63. ARRESTIN TRANSLOCATION IN ROD PHOTORECEPTORS .......... 455
W. Clay Smith, James J. Peterson, Wilda Orisme, and Astra Dinculescu

64. BINDING OF N-RETINYLIDENE-PO TO ABCA4 AND A MODEL FOR ITS TRANSPORT ACROSS MEMBRANES ......................... 465
Robert S. Molday, Seelochan Beharry, Jinhi Ahn, and Ming Zhong

65. THE CHAPERONE FUNCTION OF THE LCA PROTEIN AIPL1. AIPL1 chaperone function ............................................... 471
Jacqueline van der Spuy and Michael E. Cheetham

66. CRALBP LIGAND AND PROTEIN INTERACTIONS ..................... 477
Zhiping Wu, Sanjoy K. Bhattacharya, Zhaoyan Jin, Vera L. Bonilha, Tianyun Liu, Maria Nawrot, David C. Teller, John C. Saari, and John W. Crabb

67. FUNCTIONAL STUDY OF PHOTORECEPTOR PDEβ ........................ 485
Houbin Zhang, Jeanne M. Frederick, and Wolfgang Baehr
68. LOCALIZATION OF THE INSULIN RECEPTOR AND PHOSPHOINOSITIDE 3-KINASE IN DETERGENT-RESISTANT MEMBRANE RAFTS OF ROD PHOTORECEPTOR OUTER SEGMENTS ................................................................. 491
Raju V.S. Rajala, Michael H. Elliott, Mark E. McClellan, and Robert E. Anderson

69. MERTK ACTIVATION DURING RPE PHAGOCYTOSIS IN VIVO REQUIRES αvβ5 INTEGRIN ................................................................. 499
Silvia C. Finnemann and Emeline F. Nandrot

70. PHOTORECEPTOR RETINOL DEHYDROGENASES. An attempt to characterize the function of Rdh11 ........................................ 505
Anne Kasus-Jacobi, David G. Birch, and Robert E. Anderson

71. PIGMENT EPITHELIUM-DERIVED GROWTH FACTOR INHIBITS FETAL BOVINE SERUM STIMULATED VASCULAR ENDOThelial GROWTH FACTOR SYNTHESIS IN CULTURED HUMAN RETINAL PIGMENT EPITHELIAL CELLS ................................................................. 513
Piyush C. Kothary, Rhonda Lahiri, Lynn Kee, Nitin Sharma, Eugene Chun, Angela Kuznia, and Monte A. Del Monte

72. THE RETINAL PIGMENT EPITHELIUM APICAL MICROVILLI AND RETINAL FUNCTION ................................................................. 519
Vera L. Bonilha, Mary E. Rayborn, Sanjoy K. Bhattacharya, Xiarong Gu, John S. Crabb, John W. Crabb, and Joe G. Hollyfield

73. UPREGULATION OF TRANSGLUTAMINASE IN THE GOLDFISH RETINA DURING OPTIC NERVE REGENERATION .................. 525
Kayo Sugitani, Toru Matsukawa, Ari Maeda, and Satoru Kato

74. SURVIVAL SIGNALING IN RETINAL PIGMENT EPITHELIAL CELLS IN RESPONSE TO OXIDATIVE STRESS. Significance in retinal degenerations ................................................................. 531
Nicolas Bazan

INDEX ................................................................. 541
Retinal Degenerative Diseases
Hollyfield, J.G.; Anderson, R.E.; LaVail, M.M. (Eds.)
2006, XXII, 557 p., Hardcover
ISBN: 978-0-387-28464-4