

Sid Gilman

---

# NEUROBIOLOGY *of DISEASE*



# Contents

Contributors	ix	
Editors	xvi	
Preface	xvii	
<b>PART A. CENTRAL NERVOUS SYSTEM</b>		
<b>Section I. Metabolic Diseases</b>		
<b>Hugo W. Moser, MD</b>		
1. Lysosomal Disorders of the Nervous System Steven U. Walkley, DVM, PhD	1	
2. Neurobiology of Peroxisomal Disorders James M. Powers, MD	19	
3. Creatine Deficiency Syndromes Peter W. Schütz, MD, and Sylvia Stöckler, MD	33	
4. Leukodystrophies Shoji Tsuji	43	
<b>Section II. Neurodegenerative Disorders</b>		
<b>Gregor K. Wenning, MD, PhD</b>		
5. Parkinson's Disease Néstor Gálvez-Jiménez, MD, MSc, MHSA, FACP	51	
6. Alzheimer's Disease Kurt A. Jellinger, MD	69	
7. Multiple System Atrophy Carlo Colosimo, MD; Felix Geser, MD, PhD; Eduardo E. Benarroch, MD, DSci; and Gregor K. Wenning, MD, PhD	83	
8. Olivopontocerebellar Atrophy (OPCA) José Berciano	95	
9. Neurobiology of Progressive Supranuclear Palsy Pratap Chand, DM, FRCP, and Irene Litvan, MD	105	
<b>Section III. Genetic Diseases</b>		
<b>Christopher Gomez, MD, PhD</b>		
10. Protein Aggregation Disorders Tiago Fleming Outeiro, PhD; Pamela J. McLean, PhD; and Bradley T. Hyman, MD, PhD	111	
11. RNA-Based Disorders of Muscle and Brain Paul J. Hagerman, MD, PhD	125	
12. Ion Channel Disorders Alan L. Goldin, MD, PhD	135	
13. Spinocerebellar Ataxia Type 1 Harry T. Orr, PhD	149	
14. Mitochondrial Genetic Diseases Alice Wong, PhD, and Gino Cortopassi, PhD	157	
<b>Section IV. Neuroimmunological Disorders</b>		
<b>Anthony T. Reder, MD</b>		
15. Paraneoplastic Neurological Disorders Steven Vernino, MD, PhD, and Josep Dalmau, MD, PhD	163	
16. Systemic Lupus Erythematosus: Descriptive Past and Mechanistic Future Czesława Kowal, PhD; Cynthia Aranow, MD; Meggan Mackay, MD; Betty Diamond, MD; and Bruce T. Volpe, MD	171	
17. Progressive Multifocal Leukoencephalopathy Benjamin Brooks, MD	185	
18. Immunopathogenesis of Multiple Sclerosis Bernhard Hemmer, MD; Nicole Töpfner; and Hans-Peter Hartung, MD	197	
19. Immune-Mediated Neuropathies Michael Schroeter, MD; Bernd C. Kieseier, MD; Hubertus Köller, MD; and Hans-Peter Hartung, MD	205	
20. Hashimoto Encephalopathy Ji Y. Chong, MD	217	

<b>Section V. Cerebrovascular Diseases</b>		
<b>Lewis B. Morgenstern, MD</b>		
21. Vascular Cognitive Impairment Velandal Srikanth, PhD; Michael M. Saling, PhD; and Amanda G. Thrift, PhD	223	381
22. Cardioembolism Karen Furie, MD, MPH	235	395
23. Clinical and Neurobiological Aspects of Stroke Recovery Bruce T. Volpe, MD, and Rajiv R. Ratan, MD, PhD	241	413
24. Nonatherosclerotic Cerebral Vasculopathies Rima M. Dafer, MD, MPH, and Jose Biller, MD	255	425
25. Subarachnoid Hemorrhage David Palestrant, MD, and E. Sander Connolly, Jr., MD	265	433
26. Cerebral Ischemia: Molecular Mechanisms and Protective Therapies Kenneth R. Wagner, PhD	271	445
27. Intracerebral Hemorrhage and Intraventricular Hemorrhage–Induced Brain Injury Richard F. Keep, PhD; Guohua Xi, MD; and Julian T. Hoff, MD	281	453
<b>Section VI. Paroxysmal Disorders</b>		
<b>Timothy A. Pedley, MD</b>		
28. Idiopathic Generalized Epilepsy Jose-Luis Perez-Velasquez, PhD; Edward H. Bertram, III, MD; and O. Carter Snead, III, MD	289	473
29. Paroxysmal Dyskinesia Laurent Vercueil, MD, PhD; Anne de Saint Martin, MD; and Edouard Hirsch, MD	297	485
30. Myoclonus Pietro Mazzoni, MD, PhD, and Samay Jain, MD	305	
31. Channelopathies of the Nervous System James C. Cleland, MBChB, and Robert C. Griggs, MD	319	
32. Migraine as a Cerebral Ionopathy with Abnormal Central Sensory Processing Michel D. Ferrari, MD, PhD, and Peter J. Goadsby, MD, PhD, DSc	333	
33. Temporal Lobe Epilepsy Helen E. Scharfman, PhD, and Timothy A. Pedley, MD	349	
<b>Section VII. Neoplastic Diseases</b>		
<b>John J. Laterra, MD, PhD</b>		
34. Central Nervous System Metastases Robert J. Weil, MD, FCAS	371	537
<b>Section VIII. Infectious Diseases</b>		
<b>Richard T. Johnson, MD, FRCP</b>		
35. Meningioma Katrín Lamszus, MD; Christian Hagel, MD; and Manfred Westphal, MD		
36. Primary Central Nervous System Lymphoma Maciej Mrugala, MD, PhD, MPH; Anne Newcomer; and Tracy Batchelor, MD, MPH		
37. Neurofibromatosis 1 Linda Piersall, MS, and David H. Gutmann, MD, PhD		
38. Medulloblastoma and Primitive Neuroectodermal Tumors Said Elshihabi, MD, and James T. Rutka, MD, PhD		
39. Glioma Anders I. Persson, PhD; QiWen Fan, PhD; Joanna J. Phillips, PhD; and William A. Weiss, PhD		
<b>Section IX. Motor Neuron Diseases</b>		
<b>Eva L. Feldman, MD, PhD</b>		
40. Bacterial and Fungal Infections of the Nervous System Thomas P. Bleck, MD, FCCM		
41. Parasitic Infections Ana-Claire Meyer, MD, and Gretchen L. Birbeck, MD, MPH		
42. Prion Diseases Adriano Aguzzi, MD, PhD, DVM, hc, FRCP, FRCPPath; Frank L. Heppner, MD; Ingo M. Westner, MD; and Markus Glatzel, MD		
43. Central Nervous System Viral Infections: Clinical Aspects and Pathogenic Mechanisms Christopher Power, MD, FRCR (C), and Farshid Noorbakhsh, MD, PhD		
44. Spinal Muscular Atrophy Charlotte J. Sumner, MD, and Kenneth H. Fischbeck, MD		501
45. Amyotrophic Lateral Sclerosis–Like Syndromes Associated with Malignancy Zachary Simmons, MD		513
46. Amyotrophic Lateral Sclerosis: Idiopathic and Inherited Nicholas J. Maragakis, MD, and Jeffrey D. Rothstein, MD, PhD		521
47. Hereditary Spastic Paraparesis and Primary Lateral Sclerosis Philip A. Wilkinson, BSc, MBBS, MRCP, and Michael Swash, MD, FRCR, FRCPath		

<p>48. Poliomyelitis Eric J. Sorenson, MD</p> <p>49. Spinobulbar Muscular Atrophy (Kennedy's Disease) Jeffrey D. Zajac, MBBS, PhD, FRACP, and Karen J. Greenland, BSc, (Hons) PhD</p> <p><b>Section X. Malformations and Developmental Disorders</b> <b>Michael V. Johnston, MD</b></p> <p>50. Neurobiology of Genetic Mental Retardation Walter E. Kaufmann, MD; John C. Carter; Irena Bukelis; and David N. Lieberman, MD, PhD</p> <p>51. Cerebral Palsy Alexander H. Hoon, Jr., MD, MPH</p> <p>52. Autism Martha R. Herbert, MD, PhD</p> <p>53. Neurobiology of Dyslexia Sheryl L. Rimrodt, MD, and Laurie E. Cutting, PhD</p> <p>54. Neonatal Brain Injuries Steven P. Miller, MD, MAS, and Donna M. Ferriero, MD, MS</p> <p>55. Spina Bifida Stephen L. Kinsman, MD</p> <p>56. Circuits to Synapses: The Pathophysiology of Tourette Syndrome Harvey S. Singer, MD, and Kendra Harris, MSc</p> <p>57. Attention-Deficit Hyperactivity Disorder Paul H. Lipkin, MD, and Stewart Mostofsky, MD</p> <p>58. Congenital Hydrocephalus Stephen L. Kinsman, MD</p> <p><b>Section XI. Neurologic Manifestations of Medical Diseases</b> <b>John J. Caronna, MD</b></p> <p>59. Neurological Manifestations of Hematological Disease Babette B. Weksler, MD</p> <p>60. Neurological Manifestations of Renal Disease Ajay K. Singh, MB, MRCP, and Radhika Bhatia, MD</p> <p>61. Diabetes and Endocrine Disorders Rodica Pop-Busui, MD, PhD; Zachary London, MD; and Aaron Kellogg, BS</p> <p>62. Mechanisms and Consequences of Central Nervous System Hypoxia Meredith L. Turetz, MD, and Ronald G. Crystal, MD</p>	<p>545</p> <p>553</p> <p><b>Section XII. Sleep Disorders</b> <b>Emmanuel Mignot, MD, PhD</b></p> <p>65. REM Sleep Behavior Disorder Carlos H. Schenck, MD, and Mark W. Mahowald, MD</p> <p>66. Neurobiology of Narcolepsy and Hypersomnia Emmanuel Mignot, MD, PhD, and Jamie M. Zeitzer, PhD</p> <p>67. Restless Legs Syndrome and Periodic Limb Movements in Sleep Richard P. Allen, PhD</p> <p>68. Neurobiology of Insomnia Michael L. Perlis, PhD; Wilfred R. Pigeon, PhD; and Sean Patrick Andrews Drummond, PhD</p> <p><b>Section XIII. Substance Abuse and Basic Toxicology</b> <b>John C. M. Brust, MD</b></p> <p>69. Organic Chemicals John L. O'Donoghue, VMD, PhD, DABT</p> <p>70. Metals Luigi Manzo, MD</p> <p>71. Neurobiology of Drug Addiction Timothy P. Condon, PhD, and Curtis W. Balmer, PhD</p> <p><b>Section XIV. Imaging the Nervous System</b> <b>John C. Mazziotta, MD, PhD</b></p> <p>72. Assessment of Neurobiological Diseases with Magnetic Resonance Spectroscopy Jeffry R. Alger, PhD</p> <p>73. Magnetic Resonance Imaging John A. Detre, MD</p> <p>74. Neurovascular Computed Tomography Angiography Stuart R. Pomerantz, MD, and Michael H. Lev, MD</p> <p>75. PET Imaging in Parkinson's Disease and Other Neurodegenerative Disorders Vijay Dhawan, PhD, and David Eidelberg, MD</p>
	<p>689</p> <p>703</p> <p>709</p> <p>715</p> <p>723</p> <p>735</p> <p>745</p> <p>759</p> <p>771</p> <p>781</p> <p>793</p> <p>801</p> <p>821</p>

76. Single-Photon Emission Computed Tomography Joseph C. Masdeu, MD, PhD	829	85. Myasthenia Gravis and Myasthenic Syndromes Angela Vincent, MA (MB, BS, MSc Lond.), FRCPath, FMedSci	935
77. Functional Magnetic Resonance Imaging Gereon R. Fink, MD, PhD	839	86. Metabolic Myopathies Michio Hirano, MD	947
<b>PART B. PERIPHERAL NERVOUS SYSTEM</b>			
<b>Section XV. Peripheral Neuropathies</b> <b>James W. Russell, MD</b>			
78. Impaired Glucose Regulation and Neuropathy James W. Russell, MD, MS, FRCP; A. G. Smith, MD; and J. R. Singleton, MD	849	88. Central Autonomic Network Peter Novak, MD, PhD	969
79. Acquired Inflammatory Demyelinating and Axonal Neuropathies James W. Teener, MD, and James W. Albers, MD, PhD	859	89. Autonomic Neuropathies Steven Vernino, MD, PhD, and Phillip A. Low, MD	979
80. Toxic and Drug-Induced Neuropathies Guido Cavaletti, MD	871	90. Thermoregulation and Its Disorders Robert D. Fealey, MD	987
81. Inherited Peripheral Neuropathies Angelo Schenone, MD, and Lucilla Nobbio, PhD	885	91. Control of Blood Pressure—Normal and Abnormal Michael J. Joyner, MD; William G. Schrage, PhD; and John H. Eisenach, MD	997
82. Neurological Manifestations of Vasculitis Safwan S. Jaradeh, MD	901	<b>Section XVII. Autonomic Disorders</b> <b>Phillip A. Low, MD</b>	
83. Neuropathies Associated with Infections Paul Twydell, DO, and David N. Herrmann, MBBCh	913	88. Central Autonomic Network Peter Novak, MD, PhD	969
<b>Section XVI. Myopathies and Neuromuscular Junction Disorders</b> <b>Charles A. Thornton, MD</b>			
84. Muscular Dystrophies Leland E. Lim, MD, PhD; Charles A. Thornton, MD; and Thomas A. Rando, MD, PhD	925	89. Autonomic Neuropathies Steven Vernino, MD, PhD, and Phillip A. Low, MD	979
		90. Thermoregulation and Its Disorders Robert D. Fealey, MD	987
		91. Control of Blood Pressure—Normal and Abnormal Michael J. Joyner, MD; William G. Schrage, PhD; and John H. Eisenach, MD	997
		<b>Section XVIII. Pain</b> <b>Kenneth L. Casey, MD</b>	
		92. Neoplasm-Induced Pain Sebastiano Mercadante, MD	1007
		93. Pain Associated with the Autonomic Nervous System Wilfrid Jähnig	1021
		94. Postherpetic Neuralgia Misha-Miroslav Backonja, MD	1031
		95. Central Post-Stroke Pain David Bowsher, MD, ScD, PhD, FRCPEd, FRCPath	1039
		Index	1047