
NEURODYSTROPHIES AND NEUROLIPIDOSES

Editors

PIERRE J. VINKEN GEORGE W. BRUYN

Volume Editor

HUGO W. MOSER

REVISED SERIES 22



ELSEVIER

AMSTERDAM · LAUSANNE · NEW YORK · OXFORD · SHANNON · TOKYO

© 1996 ELSEVIER SCIENCE B.V. ALL RIGHTS RESERVED

No part of this publication may be reproduced, stored in a retrieval system or transmitted in any form or by any means, electronic, mechanical, photocopying, recording or otherwise, without the prior written permission of the publishers, Elsevier Science B.V., Copyright and Permissions Department, P.O. Box 521, 1000 AM Amsterdam, The Netherlands.

No responsibility is assumed by the publisher for any injury and/or damage to persons or property as a matter of products liability, negligence or otherwise, or from any use or operation of any methods, products, instructions or ideas contained in the material herein. Because of rapid advances in the medical sciences, the publisher recommends that independent verification of diagnoses and drug dosages should be made.

Special regulations for readers in the USA — This publication has been registered with the Copyright Clearance Center Inc., (CCC), 222 Rosewood Drive, Danvers, MA 01923. Information can be obtained from the CCC about conditions under which photocopies of parts of this publication may be made in the USA. All other copyright questions, including photocopying outside of the USA, should be referred to the publisher.



ISBN for the complete series: 0 444 90404 2

ISBN for this volume: 0 444 81285 7

ELSEVIER SCIENCE B.V.

P.O. BOX 211

1000 AE AMSTERDAM

THE NETHERLANDS

PRINTED ON ACID-FREE PAPER

PRINTED IN THE NETHERLANDS

Contents

Foreword		v
List of contributors		ix
Chapter 1.	<i>A neuropathologic overview of the neurodystrophies and neurolipidoses – J.M. Powers</i>	1
Chapter 2.	<i>Biochemistry of lipids – P. Morell and A.D. Toews</i>	33
Chapter 3.	<i>Enzymology – I. Mononen</i>	51
Chapter 4.	<i>Molecular biology – S. Byravan and A.T. Campagnoni</i>	69
Chapter 5.	<i>Bone marrow transplantation treatment for globoid cell leukodystrophy, metachromatic leukodystrophy, adrenoleukodystrophy, and Hurler syndrome – W. Krivit, E.G. Shapiro, L.A. Lockman, M. Balthazor, J. Wagner, C. Peters, J. Anderson and D. Loes</i>	87
Chapter 6.	<i>Metabolic manipulation therapy of the neurolipidoses – G.A. Grabowski and N.D. Leslie</i>	107
Chapter 7.	<i>Prospects for gene therapy in the lysosomal storage disorders – E.M. Kaye</i>	113
Chapter 8.	<i>Gaucher disease – R.O. Brady</i>	123
Chapter 9.	<i>Niemann-Pick diseases – M.T. Vanier and K. Suzuki</i>	133
Chapter 10.	<i>Metachromatic leukodystrophies – M. Philippart</i>	163
Chapter 11.	<i>Globoid leukodystrophy – E.H. Kolodny</i>	187
Chapter 12.	<i>Farber disease (lipogranulomatosis) – H.W. Moser</i>	211

Chapter 13.	<i>Fabry disease: α-galactosidase A deficiency</i> – C.M. Eng and R.J. Desnick	225
Chapter 14.	<i>The gangliosidoses</i> – K. Suzuki and K. Suzuki	247
Chapter 15.	<i>The mucopolysaccharidoses</i> – C.B. Whitley	281
Chapter 16.	<i>The oligosaccharidoses: mannosidosis, fucosidosis and aspartylglucosaminuria</i> – G. Dawson	329
Chapter 17.	<i>Schindler disease: deficient α-N-acetylgalactosaminidase activity</i> – R.J. Desnick and D. Schindler	339
Chapter 18.	<i>Sialidoses</i> – W.A. Gahl, D.M. Krasnewich and J.C. Williams	353
Chapter 19.	<i>The mucopolidoses (including I-cell disease)</i> – J.G. Leroy	377
Chapter 20.	<i>Mitochondrial disorders</i> – D.C. De Vivo, M. Hirano and S. DiMauro	389
Chapter 21.	<i>X-linked adrenoleukodystrophy</i> – P. Aubourg	447
Chapter 22.	<i>Heredopathia atactica polyneuritiformis (Refsum's disease)</i> – O.H. Skjeldal	485
Chapter 23.	<i>Generalized peroxisomal disorders and disorders of peroxisomal fatty acid oxidation</i> – R.J.A. Wanders, H.S.A. Heymans, R.B.H. Schutgens and P.G. Barth	505
Chapter 24.	<i>Rhizomelic chondrodysplasia punctata</i> – H.S.A. Heymans and R.J.A. Wanders	525
Chapter 25.	<i>Disorders of lipoprotein metabolism</i> – S.D. Kafonek and H.W. Moser	535
Chapter 26.	<i>Pelizaeus-Merzbacher disease</i> – F. Seitelberger, S. Urbanits and K.-A. Nave	559
Chapter 27.	<i>Smith-Lemli-Opitz syndrome</i> – R.I. Kelley	581
Chapter 28.	<i>Cerebrotendinous xanthomatosis</i> – A. Federico and M.T. Dotti	599
Chapter 29.	<i>Sjögren-Larsson syndrome</i> – W.B. Rizzo	615
Chapter 30.	<i>Carbohydrate-deficient glycoprotein syndromes</i> – B.A. Hagberg, G. Blennow, B. Kristiansson and H. Stibler	623

Chapter 31.	<i>Disorders of organic acid metabolism</i> – G.F. Hoffmann and K.M. Gibson	639
Chapter 32.	<i>Canavan disease</i> – R. Matalon, K. Michals and R. Kaul	661
Chapter 33.	<i>Batten disease or neuronal ceroid lipofuscinosis</i> – R.-M. Boustany	671
Chapter 34.	<i>Alexander disease</i> – A.B. Johnson	701
Chapter 35.	<i>Hallervorden-Spatz syndrome</i> – K.F. Swaiman	711
Chapter 36.	<i>Leukoencephalopathies due to exogenous factors with features of leukodystrophy: central nervous system damage caused by exposure to solvent vapors (toluene)</i> – M. Kornfeld	721
	Index	735