M.S. van der Knaap, J. Valk

Magnetic Resonance of Myelination and Myelin Disorders

Third Edition

Marjo S. van der Knaap
Jaap Valk

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With 647 Figures in 3873 parts

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Third Edition
ISBN-10 3-540-22286-3 Springer Berlin Heidelberg New York
ISBN-13 978-3-540-22286-6 Springer Berlin Heidelberg New York

Second Edition
ISBN 3-540-59277-6 Springer Berlin Heidelberg New York

Library of Congress Control Number: 2004117334

Springer is a part of Springer Science+Business Media
springeronline.com

Printed in Germany

Editor: Dr. Ute Heilmann, Heidelberg, Germany
Desk editor: Dörthe Mennecke-Bühler, Heidelberg, Germany
Production: PRO EDIT GmbH, Heidelberg, Germany
Cover-Design: Frido Steinen-Broo, Pau, Spain
Typesetting and Reproduction: AM-productions GmbH, Wiesloch, Germany
Printing and Binding: Stürtz GmbH, Würzburg, Germany

Printed on acid-free paper 21/3151Di - 5 4 3 2 1 0

Preface

Preface to the Third Edition

Reading through the prefaces of the two previous editions, we can say that much of what was said there still holds. At the same time, however, much has changed. There has been immense progress in the technical possibilities of magnetic resonance and in the knowledge of genetic defects, biochemical abnormalities, and cellular processes underlying myelin disorders. This immense progress has prompted us to embark upon the enormous task of rewriting the previous edition and adding 40 chapters. In doing so we have tried to cover most white matter disorders, hereditary and acquired, and to present a collection of images to illustrate the field to the fullest possible extent. This edition will therefore be more complete than the previous ones. The number of illustrations has increased considerably. This was necessary to reflect not only the typical patterns of a disease, but to show also the variability that exists in some disorders. The best example of this is found in Alexander disease. Genetic verification now makes it possible to recognize very different patterns of imaging abnormalities, all related to a defect in the same gene. Today's increased insight into disease classification based on increased knowledge of related genes and proteins is best reflected in the chapter on congenital muscular dystrophies.

This is the first time that we have invited a number of experts in special fields to write or co-write a chapter, in order to assure the highest level of scientific accuracy. To assemble the knowledge presented in this work we have also harvested the literature, profiting from the work and discoveries of many others.

Our thanks go to our colleagues at the VU University Medical Center and to those in other hospitals who referred their patients to us. We are indebted to all colleagues who allowed us to use their MR images, published or unpublished, making it possible for us to present illustrations of nearly all known white matter disorders. Two colleagues were particularly helpful and provided us with essential and unpublished figures: our friends Susan Blaser, from the Hospital for Sick Children in Toronto, and Zoltán Patay, from the King Faisal Hospital in Riyadh.

Many people at the VU University Medical Center have been of great technical help to us in producing high quality images and in providing secretarial assistance. The contributions of these people are mentioned separately in the acknowledgements.

Our special thanks go to patients with white matter disorders and their families. They came to see us and were willing to work with us and to go through the procedure of diagnostic testing, including MR examinations. Many patients and families were also willing to participate in our research projects to advance the understanding of white matter disorders. Patients with white matter disorders are the focus of our work. They are our most important collaborators. Often they are children. To show our gratitude to them, we have decided that all profits of this book will go to the Foundation for Children with White Matter Disorders.

Amsterdam, May 2005

M.S. van der Knaap
J. Valk

Preface to the Second Edition

The first edition of this book was well received by readers and reviewers and we are very grateful for the positive reactions. We were convinced then, and even more now, that MRI and MRS have much to offer in diagnosis, therapy monitoring and research of hereditary and acquired myelin disorders.

In the last few years, a great deal of new information has become available concerning the genetic basis of inborn errors of metabolism and neurodegenerative disorders, the role of subcellular structures, the enzyme biochemistry, the pathophysiological mechanisms of posthypoxic-ischemic cerebral damage, and the inflammatory processes in infectious and inflammatory disorders. MR images of many rare disorders have become available, either in our own experience or published by other groups. MR spectroscopy could confirm its role in certain clinical applications. Because of these developments, it was necessary for us to rewrite the book almost completely. In some fields developments are so fast that we have not have caught all the latest developments. The pattern of the new approaches has, however, been established, making the assimilation of newly available information easy.

We are extremely grateful for the help of colleagues to make this book as complete as possible. The positive reactions of those from whom we requested MR pictures or other forms of support were of enormous encouragement to us during our efforts to complete this project.

We hope this work will be as warmly welcomed by our colleagues as the first edition.

Amsterdam, January 1995

M.S. van der Knaap
J. Valk

Preface to the First Edition

Magnetic resonance imaging (MRI) is now considered to be the imaging modality of choice for the majority of disorders affecting the central nervous system. This is particularly true for gray and white matter disorders, thanks to the superb soft tissue contrast in MRI which allows gray matter, unmyelinated, and myelinated white matter to be distinguished and their respective disorders identified. The present book is devoted to the disorders of myelin and myelination. A growing amount of detailed in vivo information about myelin, myelination, and myelin disorders has been derived both from MRI and from MR spectroscopy (MRS). This prompted us to review the clinical, laboratory, biochemical, and pathological data on this subject in order to integrate all available information and to provide improved insights into normal and disordered myelin and myelination. We will show how the synthesis of all available information contributes to the interpretation of MR images.

Following a brief historical review of the increasing knowledge on myelin and myelin disorders, we propose a new classification of myelin disorders based on the subcellular localization of the enzymatic defects as far as the inborn errors of metabolism are concerned. This classification serves as a guide throughout the book. All items of the classification will be discussed and, whenever relevant and possible, illustrated by MR images.

We are aware of the fact that in a number of myelin disorders MRI is not a part of the usual diagnostic work up because a definite diagnosis is reached by other means, such as biochemical investigations of blood and urine, enzyme assessment or detection of specific antibodies. However, in many disorders MRI may facilitate a rapid diagnosis and early instigation of treatment, thus preventing structural cerebral damage. In other cases the role of MRI is to visualize the extent of brain damage and give an indication of the prognosis. In disorders which present in a nonspecific way, for instance with behavioral problems or learning difficulties, MRI can be one of the first-line investigations. It is important to be acquainted with the various MRI patterns of the myelin disorders, as an early diagnosis may be of major importance in young families with a view to the provision of adequate genetic counseling.

MRS has been of limited clinical importance until now, and its application in patients only has a short history. We do, however, expect it to be a promising technique in the field of myelin and myelin disorders in clinical as well as in basic, experimental research and have, therefore, devoted a separate chapter to this subject.

This volume was written by a neuroradiologist and a neurologist/child neurologist. It is the product of close cooperation, animated discussions, strong arguments, restructuring, rewriting, and editing, in which they had an equal share. If the reader finds value in this monograph, it is because of this dual effort.

Amsterdam and Utrecht, March 1989

J. Valk
M.S. van der Knaap

Acknowledgements

The preparation of this book was a project of several years and could not have been concluded successfully without the support and collaboration of many people. Thanks to all.

Special thanks go to our colleagues: Jeroen Vermeulen and Leo Smit, pediatric neurologists, and Frederik Barkhof and Jonas Castelijns, neuroradiologists, at the VU University Medical Center (VUMC) in Amsterdam; Martin Heitbrink and Bart Wiarda, radiologists at the Medical Center Alkmaar; and Erik Veldhuizen, radiologist at the MRI center, Amsterdam, for their continuous support during this endeavor. We are grateful to the MRI technicians at the VUMC, who guaranteed the quality of the MR examinations and had the patience and empathy to deal with very sick children and their parents. We want to mention especially the help of Karin Barbiers and Erwin Kist, who headed this team and carried out the retrieval of older examinations to the Image Management System.

We received great support from the audiovisual center at the VUMC. We are especially indebted to Daan van Eijndhoven, Rene den Engelsman, and Annuska Houtappels, who digitized older films and helped us improve the quality of the images.

Excellent secretarial help was provided by Sigrid Bruinsma, who single-handedly took care of the reference section. Staff members of the VUMC Library Els van Deventer, Linda Glas, Margreet Bosshardt, and Cisca Frederiks were very helpful in providing us with the necessary literature.

Technical support and guidance with computer programs and settings were provided by the Department of Informatics of the VUMC. We are grateful for their kind and prompt assistance. Special thanks go to Michiel Sprenger, Guido Zonneveld, and Peter Theijsmeijer.

We acknowledge the continuous friendly and encouraging support of the editorial staff of Springer-Verlag, Dr. Ute Heilmann and Mrs Dörthe Menneke-Bühler.

Amsterdam, May 2005

M.S. van der Knaap
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List of Abbreviations

ACE	angiotensin converting enzyme
ACTH	adrenocorticotropic hormone
AD	Alexander disease
ADC	apparent diffusion coefficient
ADEM	acute disseminated encephalomyelitis
ADP	adenosine diphosphate
AD PEO	autosomal dominant progressive external ophthalmoplegia
AHEM	acute hemorrhagic encephalomyelitis
AIDS	acquired immunodeficiency syndrome
ALD	adrenoleukodystrophy
ALDP	ALD protein
ALL	acute lymphocytic leukemia
AMN	adrenomyeloneuropathy
ANCAs	anti-neutrophil cytoplasm antibodies
ANCL	adult neuronal ceroid lipofuscinosis (or Kufs disease)
AP_4	2-amino-4-phosphonobutyrate
APLA	anti-phospholipid antibodies
APBD	adult polyglucosan body disease
apoE	apolipoprotein E
APP	amyloid precursor protein
aPTT	activated partial thromboplastin time
ASLD	argininosuccinate lyase deficiency
ASSD	argininosuccinate synthetase deficiency
ATP	adenosine triphosphate
BAEP	brain stem auditory evoked potential
BCNU	*bis*-chloroethyl-nitrosourea
BDNF	brain-derived neurotrophic factor
bFGF	basic fibroblast growth factor
BIDS	brittle hair, impaired intelligence, decreased fertility, short stature (syndrome)
BMAA	β-*N*-methylamino-L-alanine
BOMAA	β-*N*-oxalylmethylamino-L-alanine
BPD	D-bifunctional protein deficiency
CAA	cerebral amyloid angiopathy
CACH	childhood ataxia with central nervous system hypomyelination
CACT	mitochondrial carnitine/acylcarnitine transporter
CADASIL	cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
cANCA	cytoplasmic form of ANCA
CARASIL	cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
CAMFAK	cataracts–microcephaly–failure to thrive–kyphoscoliosis (syndrome)
CD	Canavan disease; cluster determinant
Cho	choline
CIPO	chronic intestinal pseudo-obstruction
CIS	clinically isolated symptom
CK	creatine kinase
CMD	congenital muscular dystrophy
CMT	Charcot–Marie–Tooth disease
CMTX	X-linked form of CMT
CMV	cytomegalovirus
CNP	2'3'-cyclic nucleotide 3'-phosphodiesterase
CNS	central nervous system
COFS	cerebro-oculofacioskeletal (syndrome)
COX	cytochrome-*c* oxidase
CPEO	chronic progressive external ophthalmoplegia
CPM	central pontine myelinolysis
CPSD	carbamyl phosphate synthetase deficiency
CPT	carnitine palmitoyl transferase
Cr	creatine
CREST	*c*alcinosis, *R*aynaud syndrome, *e*sophageal problems, *s*clerodactylia, and *t*elangiectasia (syndrome)
CS	Cockayne syndrome; concentric sclerosis (or Baló disease)
CSF	cerebrospinal fluid
CSI	chemical shift imaging
CT	computed tomography/tomogram
CTX	cerebrotendinous xanthomatosis
DAB	diaminobenzidine
DAGC	dystrophin-associated glycoprotein complex
DAI	diffuse axonal injury
DAVF	cranial dural arteriovenous fistula
DHAPAT	dihydroxyacetonephosphate acyltransferase
DM 1	myotonic dystrophy type 1
DM 2	myotonic dystrophy type 2
DNA	deoxyribonucleic acid
DNC	deoxynucleotide carrier
dNTP	deoxyribonucleoside triphosphate
DOA	dominant optic atrophy
DOPA	dihydroxyphenylalanine
DPHL	delayed posthypoxic leukoencephalopathy
DRPLA	dentatorubropallidoluysian atrophy

DS	diffuse sclerosis (or Schilder disease)
DSA	digital subtraction angiography
DTI	diffusion tensor imaging
DWI	diffusion-weighted imaging
EAA	excitatory amino acid
EAE	experimental allergic encephalomyelitis
ECD	ethyl cysteinate dimer
ECG	electrocardiography/electrocardiogram
EDSS	Expanded Disability Status Scale
EEG	electroencephalogram
EGF	epidermal growth factor
eIF	eukaryotic initiation factor
ELISA	enzyme-linked immunosorbent assay
EMG	electromyogram
EPI	echo planar imaging
EPM	extrapontine myelinolysis
EPMR	progressive epilepsy with mental retardation
ERG	electroretinography/electroretinogram
FA	fractional anisotropy
FAD	flavin adenine dinucleotide
FADH2	flavin adenine dinucleotide, reduced
FCMD	Fukuyama congenital muscular dystrophy
FD	Fabry disease
FISH	fluorescent in situ hybridization
FLAIR	fluid-attenuated inversion recovery
FSE	fast spin echo
FSH	follicle-stimulating hormone
5-FU	5-fluorouracil
FvLINCL	Finnish variant of late-infantile neuronal ceroid lipofuscinosis
GA	gestational age
GABA	γ-aminobutyric acid
GAMT	guanidinoacetate methyltransferase
GAN	giant axonal neuropathy
GDP	guanosine diphosphate
GE	gradient echo
GEF	guanine-nucleotide exchange factor
GFAP	glial fibrillary acidic protein
GIP	general insertion protein
GLD	globoid cell leukodystrophy
Glx	glutamine, glutamate, GABA
GOM	granular osmiophilic material
GRACILE	growth retardation, aminoaciduria, cholestasis, iron overload, lactic acidosis, and early death (syndrome)
GROD	granular osmiophilic deposits
GTE	glyceryl trierucate
GTO	glyceryl trioleate
GTP	guanosine triphosphate
GVHD	graft-versus-host disease
HAART	highly active/aggressive anti-retroviral treatment
HABC	hypomyelination with atrophy of the basal ganglia and cerebellum
HCHWA-D	Dutch type of hereditary cerebral hemorrhage with amyloidosis
HDL	high-density lipoproteins
HDLS	hereditary diffuse leukoencephalopathy with spheroids
5HIAA	5-hydroxyindoleacetic acid
HIV-1	human immunodeficiency virus type 1
HLA	human leukocyte antigen
HMG-CoA	3-hydroxy-3-methylglutaryl-coenzyme A
HMI	hypomelanosis of Ito
HMPAO	hexamethylpropyleneamine oxime
HSP	hereditary spastic paraplegia; heat shock protein
HTLV	human T-cell lymphotropic virus
HUS	hemolytic–uremic syndrome
HVA	homovanillic acid
IBIDS	ichthyosis, brittle hair, impaired intelligence, decreased fertility, short stature (syndrome)
IFN	interferon
Ig	immunoglobulin
IGF	insulin-like growth factor
INCL	infantile neuronal ceroid lipofuscinosis (or Santavuori disease)
IP	incontinentia pigmenti
IQ	intelligence quotient
IR	inversion recovery
IRD	infantile Refsum disease
ISIS	image-selective in vivo spectroscopy
ISSD	severe infantile sialic acid storage disease
IVL	intravascular lymphomatosis
JNCL	juvenile neuronal ceroid lipofuscinosis (or Spielmeyer–Vogt disease, or Batten disease)
KA	kainate
kDa	kiloDalton
KSS	Kearns–Sayre syndrome
LAMP	lysosome-associated membrane protein
LBSL	leukoencephalopathy with brain stem and spinal cord involvement and elevated white matter lactate
LCC	leukoencephalopathy with calcifications and cysts
LCH	Langerhans cell histiocytosis
LDL	low-density lipoproteins
LGMD	limb girdle muscular dystrophy
LH	luteinizing hormone
LHON	Leber hereditary optic neuropathy
LINCL	late-infantile neuronal ceroid lipofuscinosis (or Jansky–Bielschowsky disease)
MAG	myelin-associated glycoprotein
MAP	microtubule-associated protein
MBS	Marchiafava–Bignami syndrome
MBP	myelin basic protein

MCE	multicystic encephalopathy
MD	Menkes disease; myotonic dystrophy
MDC1A	merosin-deficient congenital muscular dystrophy
MEB	muscle–eye–brain disease
MELAS	mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes
MEPOP	mitochondrial encephalomyopathy with sensorimotor polyneuropathy, ophthalmoplegia, and pseudo-obstruction
MERRF	myoclonic epilepsy with ragged red fibers
MHC	major histocompatibility complex
MHPG	3-methoxy-4-hydroxyphenylglycol
MICS	microcephaly–intracranial calcifications syndrome
MIL	multifocal inflammatory leukoencephalopathy
mIns	*myo*-inositol
MLC	megalencephalic leukoencephalopathy with subcortical cysts
MLD	metachromatic leukodystrophy
MNGIE	mitochondrial neurogastrointestinal encephalomyopathy
MOBP	myelin-associated oligodendrocytic basic protein
MOG	myelin oligodendrocyte glycoprotein
MOM	mitochondrial outer membrane
MOSP	myelin-/oligodendrocyte-specific protein
MPP	mitochondrial processing peptidase
MPS	mucopolysaccharidoses; mucopoly-saccharidoses
MPTP	methylphenyltetrahydropyridine
MR	magnetic resonance
MRA	magnetic resonance angiography
MRI	magnetic resonance imaging
mRNA	messenger RNA
MRS	magnetic resonance spectroscopy
MS	multiple sclerosis
MSD	multiple sulfatase deficiency
MSUD	maple syrup urine disease
MT	magnetization transfer
mtDNA	mitochondrial DNA
MTI	magnetization transfer imaging
MTR	magnetization transfer ratio
NAA	*N*-acetylaspartate
NAAG	*N*-acetylaspartyl glutamate
NAD	nicotinamide adenine dinucleotide
NADH	nicotinamide adenine dinucleotide, reduced
NALD	neonatal adrenoleukodystrophy
NARP	neurogenic muscle weakness, ataxia, and retinitis pigmentosa
NAWM	normal-appearing white matter
NBCA	*n*-butyl cyanoacrylate
NCL	neuronal ceroid lipofuscinosis
nDNA	nuclear DNA
NKH	nonketotic hyperglycinemia
NMDA	*N*-methyl-D-aspartate
NMO	neuromyelitis optica (or Devic disease)
NRTI	nucleoside analogue reverse transcriptase inhibitor
NT	neurotrophin
OCRL	oculocerebrorenal syndrome of Lowe
ODDD	oculodentodigital dysplasia
OGIMD	oculogastrointestinal muscular dystrophy
OHS	occipital horn syndrome
OMgp	oligodendrocyte myelin glycoprotein
ONMR	onychotrichodysplasia, neutropenia, mental retardation (syndrome)
OSP	oligodendrocyte-specific protein
OTCD	ornithine transcarbamylase deficiency
PACNS	primary angiitis of the CNS
PAF	platelet activating factor
PAN	polyarteritis nodosa
pANCA	perinuclear form of ANCA
PAS	periodic acid–Schiff
PCD	pyruvate carboxylase deficiency
PCr	phosphocreatine
PCR	polymerase chain reaction
PDE	phosphodiesters
PDGF	platelet-derived growth factor
PDHc	pyruvate dehydrogenase complex
PEP	processing enhancing protein
PET	positron emission tomography
Pi	inorganic phosphate
PIBIDS	photosensitivity, ichthyosis, brittle hair, impaired intelligence, decreased fertility, short stature (syndrome)
PIP2	phosphatidylinositol 4,5-biphosphate
PKU	phenylketonuria
PLOSL	polycystic lipomembranous osteo-dysplasia with sclerosing leuko-encephalopathy
PLP	proteolipid protein
PMD	Pelizaeus–Merzbacher disease; proximal myotonic dystrophy
PME	phosphomonoesters
PML	progressive multifocal leuko-encephalopathy
PMP	peroxisomal membrane protein
PNS	peripheral nervous system
POLD	pigmentary orthochromatic leukodystrophy
POLIP	polyneuropathy, ophthalmoplegia, leukoencephalopathy, and intestinal pseudo-obstruction
PPAR	peroxisome proliferator activating receptor
ppm	parts per million

PPRE	peroxisome proliferator response element
PPT1	palmitoyl protein thioesterase 1
PRES	posterior reversible encephalopathy syndrome
PRESS	point-resolved spectroscopy
PROMM	proximal myotonic myopathy
PTS	peroxisome targeting signals
PVA	polyvinyl alcohol
PVL	periventricular leukomalacia
QA	quisqualate
RCDP	rhizomelic chondrodysplasia punctata
RD	Refsum disease
RF	radiofrequency
RNA	ribonucleic acid
RPLS	reversible posterior leukoencephalopathy syndrome
RPR	rapid plasma reagin (test)
RR	relapsing remitting
rRNA	ribosomal RNA
RXR	retinoic acid receptor
SAE	subcortical arteriosclerotic encephalopathy
SAP	sphingolipid activator protein
SCA	spinocerebellar ataxia
SCL	subcortical leukomalacia
SD	Salla disease
SE	spine echo
SIBIDS	osteosclerosis, ichthyosis, brittle hair, impaired intelligence, decreased fertility, short stature (syndrome)
SLE	systemic lupus erythematosus
SLS	Sjögren–Larsson syndrome
SP	secondary progressive
SPECT	single photon emission computed tomography
SPG2	spastic paraparesis type 2
SSEP	somatosensory evoked potential
SSPE	subacute sclerosing panencephalitis
STEAM	stimulated-echo acquisition mode
STIR	short tau inversion recovery
T	Tesla
TE	toxic encephalopathy; echo time
TI	inversion time
TNF-α	tumor necrosis factor-alpha
TORCH	toxoplasmosis, rubella, cytomegalovirus, herpes simplex
TPP1	tripeptidyl peptidase 1
TR	repetition time
tRNA	transfer RNA
TSD	Tay–Sachs disease
TSE	turbo spin echo
TTD	trichothiodystrophy with photosensitivity
TTP	thrombotic thrombocytopenic purpura
TvLINCL	Turkish variant of late-infantile neuronal ceroid lipofuscinosis
TYROBP	TYRO protein tyrosine kinase binding protein
UDP	uridine diphosphate
US	ultrasound/ultrasonography
UV	ultraviolet
V-CAM	cellular adhesion molecules
VDAC	voltage-dependent, anion-selective channel
VDRL	Venereal Disease Research Laboratory (test)
VEGF	vascular endothelial growth factor
VEP	visual evoked potential
VLA-4	very late antigen 4
VLCFA	very-long-chain fatty acids
vLINCL	variant late-infantile neuronal ceroid lipofuscinosis
VMA	vanillyl mandelic acid
VWM	vanishing white matter
WD	Wilson disease
WM	white matter
WWS	Walker–Warburg syndrome
XALD	X-linked adrenoleukodystrophy
XP	xeroderma pigmentosum
ZS	Zellweger syndrome