Movement Disorders in Neurologic and Systemic Disease

Movement Disorders in Neurologic and Systemic Disease

Editors

Werner Poewe, MD
Department of Neurology, Innsbruck Medical University, Innsbruck, Austria

Joseph Jankovic, MD
Parkinson's Disease Center and Movement Disorders Clinic, Department of Neurology, Baylor College of Medicine, Houston, TX, USA

CAMBRIDGE UNIVERSITY PRESS

CAMBRIDGE
UNIVERSITY PRESS

University Printing House, Cambridge CB2 8BS, United Kingdom

Published in the United States of America by Cambridge University Press, New York

Cambridge University Press is part of the University of Cambridge.

It furthers the University's mission by disseminating knowledge in the pursuit of education, learning and research at the highest international levels of excellence.

www.cambridge.org
Information on this title: www.cambridge.org/9781107024618

Movement Disorders in Neurologic and Systemic Disease, ed. Werner Poewe and Joseph Jankovic. Published by Cambridge University Press.

First published 2014

Printed in the United Kingdom by Clays, St Ives plc

A catalog record for this publication is available from the British Library

Library of Congress Cataloging-in-Publication Data
Movement disorders in neurologic and systemic disease / editors, Werner Poewe, Joseph Jankovic.
p. ; cm.
Includes bibliographical references and index.
ISBN 978-1-107-02461-8 (Hardback)
I. Poewe, W., editor of compilation. II. Jankovic, Joseph, editor of compilation.
[DNLM: 1. Movement Disorders–etiology. 2. Movement Disorders–physiopathology. 3. Neurologic Manifestations. WL 390]
RC376.5
616.8′3–dc23 2013027998

ISBN 978-1-107-02461-8 Hardback

Additional resources for this publication at www.cambridge.org/9781107024618

Contents

Contents

Contributors

Marjolein B. Aerts, MD
Department of Neurology and Parkinson Centre Nijmegen, Donders Institute for Brain Cognition & Behaviour, Radboud University Nijmegen Medical Centre, Nijmegen, the Netherlands

Angelo Antonini, MD
UO Malattia Parkinson, IRCCS San Camillo Venezia e Neurologia, AO University Padova, Padova, Italy

José Fidel Baizabal-Carvallo, MD, MSc
Parkinson's Disease Center and Movement Disorders Clinic, Baylor College of Medicine, Department of Neurology, Houston, TX, USA

Kailash P. Bhatia, MD, FRCP
Sobell Department of Motor Neuroscience & Movement Disorders, UCL Institute of Neurology, London, UK

Bastiaan R. Bloem, MD, PhD
Department of Neurology and Parkinson Centre Nijmegen, Donders Institute for Brain Cognition & Behaviour, Radboud University Nijmegen Medical Centre, Nijmegen, the Netherlands

Francisco Cardoso, MD, PhD
Movement Disorders Unit, Neurology Division, Internal Medicine Department, Federal University of Minas Gerais, Belo Horizonte, MG, Brazil

Miryam Carecchio, MD
Sobell Department of Motor Neuroscience & Movement Disorders, UCL Institute of Neurology, London, UK; Department of Neurology, Amedeo Avogadro University, Novara, Italy

Leslie J. Cloud, MD, MSc
Parkinson's & Movement Disorders Center, Virginia Commonwealth University, Richmond, VA, USA

Cynthia L. Comella, MD
Department of Neurological Sciences, Rush University Medical Center, Chicago, IL, USA

Josep Dalmau, MD, PhD
Department of Neurology, University of Pennsylvania, Philadelphia, PA, USA

Sheila R. Eichenseer, MD
Department of Neurological Sciences, Rush University Medical Center, Chicago, IL, USA

Alessandra Fanciulli, MD
Department of Neurology & Psychiatry, Sapienza, Università di Roma, Rome, Italy

Emilio Fernández-Álvarez, MD
Department of Neurology, Hospital San Juan de Dios, Barcelona, Spain

Joseph M. Ferrara, MD
Division of Neurology, Virginia Tech Carilion School of Medicine & Research Institute, Roanoke, VA, USA

Joaquim J. Ferreira, MD, PhD
Neurological Clinical Research Unit, Instituto de Medicina Molecular, and Laboratory of Clinical Pharmacology & Therapeutics, Faculty of Medicine, University of Lisbon, Portugal

Birgit Frauscher, MD
Department of Neurology, Innsbruck Medical University, Innsbruck, Austria

Steven J. Frucht, MD
Movement Disorders Division, Department of Neurology, Mount Sinai School of Medicine, New York, NY, USA

Deborah A. Hall, MD, PhD
Department of Neurological Sciences, Rush University Medical Center, Chicago, IL, USA

Contributors

Taku Hatano, MD, PhD
Department of Neurology, Juntendo University, School of Medicine, Tokyo, Japan

Nobutaka Hattori, MD, PhD
Department of Neurology, Juntendo University, School of Medicine, Tokyo, Japan

Lindsay Hinkle-Johnston, DO
Department of Neurology, University of Massachusetts Medical School, Worcester, MA, USA

Joseph Jankovic, MD
Parkinson's Disease Center and Movement Disorders Clinic, Department of Neurology, Baylor College of Medicine, Houston, TX, USA

Ryuji Kaji, MD, PhD
Department of Neurology, Tokushima University, Tokushima, Japan

Sarah M. Kranick, MD
Section of Infections of the Nervous System, National Institute of Neurological Diseases & Stroke, National Institutes of Health, Bethesda, MD, USA

Shin-ichiro Kubo, MD, PhD
Department of Neurology, Juntendo University, School of Medicine, Tokyo, Japan

Jessica B. Lehosit, DO
Parkinson's & Movement Disorders Center, Virginia Commonwealth University, Richmond, VA, USA

Timothy Lynch, BSc, DCH, FRCPI, FRCP
Dublin Neurological Institute, Mater Hospital, Dublin, Ireland

Benjamin Matosevic, MD
Department of Neurology, Innsbruck Medical University, Innsbruck, Austria

Raja Mehanna, MD
Parkinson's Disease Center and Movement Disorders Clinic, Department of Neurology, Baylor College of Medicine, Houston, TX, USA

Shyamal H. Mehta, MD, PhD
Movement Disorders Program, Georgia's Health Sciences University, Augusta, GA, USA

Marcelo Merello, MD, PhD
Department of Neuroscience FLENI and Universidad Católica (UCA), Buenos Aires, Argentina

Yoshikuni Mizuno, MD, PhD
Department of Neurology, Juntendo University, School of Medicine, Tokyo; Department of Neuroregenerative Medicine, Kitasato University, School of Medicine, Kanagawa, Japan

Renato Puppi Munhoz, MD, MSc
Neurology Service, Hospital Cajuru, Pontificial Catholic University; Movement Disorders Unit, Neurology Service, Internal Medicine Department, Hospital de Clinicas, Federal University of Paraná, Curitiba, PR, Brazil

Avindra Nath, MD
Section of Infections of the Nervous System, National Institute of Neurological Diseases & Stroke, National Institutes of Health, Bethesda, MD, USA

K. Amande M. Pauls, MD, PhD
Department of Neurology, University Hospital Cologne, Cologne, Germany

Tasneem Peeraully, MBBS, BSc
Department of Neurology, Ronald Reagan UCLA Medical Center, Los Angeles, CA, USA

Werner Poewe, MD
Department of Neurology, Innsbruck Medical University, Innsbruck, Austria

Ritesh A. Ramdhani, MD
Movement Disorders Division, Department of Neurology, Mount Sinai School of Medicine, New York, NY, USA

Kinley Roberts
Dublin Neurological Institute, Mater Hospital, Dublin, Ireland

Agathe Roubertie, MD, PhD
Department of Neurology, Hôpital Gui de Chauliac, Montpellier, France

Erich Schmutzhard, MD
Department of Neurology, Innsbruck Medical University, Innsbruck, Austria

Susanne A. Schneider, MD, PhD
Department of Neurology, Christian-Albrechts-University, Kiel, Germany

Kapil D. Sethi, MD, FRCP
Movement Disorders Program, Georgia's Health Sciences University, Augusta, GA, USA

Pille Taba, MD, PhD
Department of Neurology & Neurosurgery, University of Tartu, Tartu, Estonia

Eng-King Tan, MBBS, MRCP, FRCP
Neuroscience & Behavioral Disorders Program, Duke-NUS Graduate Medical School, Singapore

Hélio A. G. Teive, MD, PhD
Movement Disorders Unit, Neurology Service, Internal Medicine Department, Hospital de Clinicas, Federal University of Paraná, Curitiba, PR, Brazil

Tiago Teodoro, MD
Neurological Clinical Research Unit, Instituto de Medicina Molecular, Lisbon, Portugal

Philip D. Thompson, PhD, FRACP
University Department of Medicine, University of Adelaide; Department of Neurology, Royal Adelaide Hospital, Adelaide, Australia

Lars Timmermann, MD
Department of Neurology, University Hospital Cologne, Cologne, Germany

Francesc Valldeoriola, MD, PhD
IDIBAPS-Hospital Clínic, Service of Neurology, University of Barcelona, Barcelona, Spain

Bart P. van de Warrenburg, MD, PhD
Department of Neurology and Parkinson Centre Nijmegen, Donders Institute for Brain Cognition & Behaviour, Radboud University Nijmegen Medical Centre, Nijmegen, the Netherlands

Gregor K. Wenning, MD, PhD, MSc
Division of Neurobiology, Department of Neurology, Medical University Innsbruck, Innsbruck, Austria

Videos

The following videos can be found at www.cambridge.org/9781107024618

Chapter 1

Parkinsonism

1.1 Parkinson's disease (untreated) manifested by hypomimia, low-volume-hypokinetic dysarthria, right-hand and jaw-rest tremor, bradykinesia, and decreased right-arm swing.

1.2 Parkinson's disease, currently on only a small dose of pramipexole, manifested by right-hand rest tremor, bradykinesia, shuffling gait, leaning to the right side (Pisa sign), and bending of his trunk (camptocormia) which is corrected in supine position.

1.3 Parkinson's disease and re-emergent tremor.

1.4 Progressive supranuclear palsy with square-wave jerks, marked limitation of downward gaze which is corrected with an oculocephalic maneuver, stiff gait with legs extended when walking, crossing feet when turning, and difficulties sitting down.

1.5 Corticobasal degeneration with severe apraxia in the right more than left hand (also exhibits right alien-hand syndrome). Her MRI of the brain shows marked atrophy, especially in the left fronto-parietal cortex.

1.6 Corticobasal degeneration with left-hand spontaneous and stimulus-induced myoclonus and marked apraxia.

Myoclonus

1.7 Myoclonus-dystonia due to mutation in the ε-sarcoglycan (SGCE), manifested by action myoclonus and dystonic writer's cramp. His daughter and son are also affected.

1.8 Myoclonus secondary to EPM1 gene mutation, also referred to as Unverricht-Lundborg disease or Baltic myoclonus.

Chorea/Ballism

1.9 Huntington's disease, with forty-three CAG repeats in the *huntingtin* gene, with a four-year history of progressive chorea, irritability, and cognitive decline. In addition to generalized chorea, which she tries to camouflage by touching her face ("parakinesia"), she also exhibits pendular and hung-up knee-jerk reflexes.

1.10 Sydenham's chorea in a 13-year-old girl manifested by sudden-onset compulsive behavior, followed by rapidly progressive chorea, initially misdiagnosed as a "psychogenic" disorder. She had high ASO titers and improved with penicillin which had to be restarted when symptoms recurred.

1.11 This 19-year-old woman with history of systemic lupus erythematosus, manifested in the past by "meningitis" at age 10 and subsequent arthralgias, myalgias, and pericarditis, presented with a ten-day history of rapidly progressive generalized chorea. The patient markedly improved with tetrabenazine.

1.12 Left hemiballism after right subcortical ischemic stroke involving the subthalamic nucleus.

Tics

1.13 Young girl with Tourette's syndrome manifested chiefly by clonic and dystonic facial and neck tics often preceded by premonitory sensations.

1.14 A 62-year-old man with life-long history of motor and phonic tics, including coprolalia.

About one year after starting ziprasidone for the tics he has developed mild oro-facial lingual stereotypy, suggestive of tardive dyskinesia.

Dystonia

1.15 Task-specific dystonia manifested by writer's cramp and hand flexion when performing equine dentistry. She also exhibits mirror dystonia with extension of the right third to fifth fingers and abduction of the fifth finger.

1.16 Patient with blepharospasm and oromandibular dystonia (cranial dystonia), markedly improved after botulinum toxin injections.

1.17 Blepharospasm with apraxia of eyelid opening.

1.18 Tardive dystonia in a schizophrenic man after five months of treatment with ziprasidone. He exhibits cervical dystonia with retrocollis and torticollis to the left, trunk dystonia with opisthotonic posturing, and arm extension and pronation.

1.19 Patient with DYT1 dystonia, manifested by generalized dystonia resulting in dromedary gait and dystonic trunk flexion (camptocormia) and severe dysathria due to jaw-opening dystonia, which markedly improved after bilateral pallidotomy.

Tremor

1.20 Classic essential tremor for at least twenty years involving chiefly her head, voice, hands, and legs improved with alcohol and worsened with caffeine. She has marked difficulties with her handwriting and other activities that require fine motor coordination. She also has slight difficulties with her balance and performing tandem gait.

Other movement disorders

1.21 Classic tardive dyskinesia occurring two weeks after discontinuing chronic treatment with prochlorperazine.

1.22 Chinese couple with left hemifacial spasm.

1.23 Stiff person syndrome with positive anti-GAD antibodies.

Chapter 6

6.1 Sydenham chorea.

6.2 HIV-related chorea.

Chapter 8

8.1 A 22-year-old male with propionic acidemia. Wide-based ataxic gait, generalized chorea with axial dystonia and superimposed jerks of upper limbs.

8.2 Supranuclear gaze palsy with predominant downgaze impairment in a 36-year-old male with adult-onset Niemann-Pick disease type C presenting with cognitive decline.

8.3 A 45-year-old male with cerebrotendinous xanthomatosis. Akinetic-rigid parkinsonism with severe bilateral bradykinesia on tapping, more marked on the right side, hypomimic face, short-stepped gait and bilateral pes cavus. Bilateral xanthomas of the Achilles tendons are visible. This patient had reduced bilateral putaminal tracer uptake on DAT scan.

Chapter 10

10.1 Illustrative case of an elderly female patient with diabetes mellitus who developed HBHC due to a contralateral basal ganglia lacunar ischemic lesion as shown in the MRI imaging in Figure 10.1.

10.2 A 60-year-old male of Japanese descent, with a one-month history of rigid akinetic parkinsonism and gait ataxia. The patient had a previous history of diabetes and chronic renal failure undergoing hemodialysis three times a week. His serum ammonia level was 66 mMol/l and creatinine level was 11 mg/dl; with arterial gasometry showing a pH of 7.27 with HCO_3 of 7 mEq/l, compatible with metabolic acidosis. Brain MRI showed symmetric hyperintense sign in the basal ganglia, identical to the lentiform fork sign on T2 and FLAIR described by Kumar and Goyal (Figure 10.2).

Chapter 12

12.1 Limb chorea and dystonia facial grimacing in a 56-year-old male with acute basal ganglia lesions in the setting of uremia. He had end-stage renal failure secondary to diabetic nephropathy and has been doing hemodialysis

for fifteen years. After adjustment of dialysis program and treatment with sulpiride, involuntary movements resolved. (See also Figures 12.1 and 12.2.)

Chapter 15

15.1 Manganism in welders.
15.2 Manganism in drug abusers.

Chapter 16

16.1 A 42-year-old drummer six weeks after "chasing the dragon."
16.2 Serotonin syndrome in a lady with a history of prothiaden and phenelzine use.

Chapter 17

17.1 A 26-year-old male with action-induced left foot dystonia (equinovarus posturing at the ankle while walking). The movement disorder had developed approximately six months following right thalamic hemorrhage.
17.2 A 78-year-old male with a wake-up stroke presenting with acute onset hemichorea on the left involving the left side of his face and L-sided extremities.
17.3 Limb-shaking attack involving right arm in a 58-year-old male patient with left internal carotid occlusion. Video was taken by the patient's wife while visiting him in hospital.
17.4 A 23-year-old female three months after left-sided thalamic hemorrhage and subacute onset of mixed movement disorder of the right hand comprising irregular myoclonic jerking and dystonic posturing with short episodes of distal hand tremor.

Chapter 19

19 PSP look-alike in a patient with primary CNS lymphoma.
Segment 1: Upon admission, the patient exhibits vertical gaze palsy, bradykinesia, and postural instability. The patient's gait was wide-based with start hesitation.
Segment 2: After chemotherapy, the patient exhibited no supranuclear gaze palsy or parkinsonism.

Chapter 20

20.1 Multidirectional opsoclonus in a 79-year-old male with severe TBE viral meningoencephalitis.
20.2 A 24-year-old female immune-competent patient with subacute onset of mixed movement disorder involving the left arm. Video shows myoclonic jerking of the fingers of her left hand plus dystonic posturing. In addition, episodes of irregular distal, postural, and kinetic arm tremor can be seen (from Stockhammer *et al.* 2000, with kind permission).
20.3 Right-sided hemichorea in a 74-year-old male with pulmonary tuberculosis and tuberculoma involving the left thalamus and subthalamic region (see Figure 20.6).
20.4 Opsoclonus in a 78-year-old woman with neuroborreliosis. Symptoms completely resolved following a two-week course of intravenous therapy with ceftriaxone.
20.5 Masticatory-facial myorhythmia in a case of Whipple's Disease (courtesy Professor J. Jankovic).
20.6 A 49-year-old male with CNS toxoplasmosis involving the basal ganglia bilaterally (see Figure 20.6) and severe akinetic-rigid parkinsonism. Note facial hypomimia, marked slowing and decrement of repetive finger, hand and foot tapping movements, L>R, as well as marked difficulty rising from a chair and shuffling, small-stepped gait.

Chapter 21

21.1 Video-polysomnography shows rhythmic movements which usually occur with a frequency of 0.5–2 Hz.
21.2 An example of a patient with bruxism.
21.3 An example of an RBD episode.
21.4 Due to RLS symptoms, patients frequently present with difficulties falling asleep or sleep maintenance insomnia.
21.5 Typical example of PLMS.

Abbreviations

3,4-MDMA	3,4-methylenedioxymethamphetamine
5MTHF	5-methyltetrahydrofolate
AADC-D	aromatic L-amino acid decarboxylase deficiency
AARS	atlanto-axial rotatory subluxation
ABL	abetalipoproteinemia
ACD	alcoholic cerebellar degeneration
ACR	American College of Rheumatology
ACTH	adrenocorticotropic hormone
ADC	apparent diffusion coefficient
ADNFLE	autosomal dominant nocturnal frontal lobe epilepsy
AED	anti-epileptic drug
AGA	anti-gliadin antibodies
AGAT	arginine glycine amidinotransferase
AHD	acquired hepatocerebral degeneration
AIDS	acquired immune deficiency syndrome
ALS	amyotrophic lateral sclerosis
AMRF	action myoclonus-renal failure
ANA	anti-nuclear antibodies
ANI	asymptomatic neurocognitive impairment
AP	atypical parkinsonism
aPL	antiphospholipid
APS	antiphospholipid syndrome
AR GTPCH-D	autosomal-recessive GTP cyclohydrolase 1 deficiency
ART	antiretroviral therapy
ARX	aristaless-related homeobox
ASO	Antistreptolysin
BBB	blood-brain barrier
BCAA	branched-chain amino acid
BCKD	branched-chain α-keto acid dehydrogenase
BD	Behçet's disease
BFNC	benign familial neonatal convulsions
BFNIS	benign familial neonatal-infantile seizures
BFNS	benign neonatal familial seizures
BHC	benign hereditary chorea
b.i.d.	twice a day
BMAA	beta-methylamino-L-alanine
BMI	body mass index
BP	blood pressure
BRBGD	biotin-responsive basal ganglia disease
BSMI	benign sleep myoclonus of infancy
BUN	blood urea nitrogen
CAE	childhood absence epilepsy
CAG	cytosine-adenine-guanine
cAMP	cyclic adenosine monophosphate
$CaNa_2EDTA$	calcium salt of ethylene diamine tetraacetic acid
CANS	childhood acute neuropsychiatric symptoms
cART	combined antiretroviral therapy
CBD	corticobasal degeneration
CBGD	corticobasal ganglionic degeneration
CD	celiac disease
CFD	cerebral folate deficiency
CGD	chronic granulomatous disease
ChAc	chorea-acanthocytosis
CK	creatine kinase
CNS	central nervous system
CP	cerebral palsy
CPAP	continuous positive airway pressure
CPK	creatine phosphokinase
CRMP5	CV2/collapsin response mediator protein-5
CRPS	complex regional pain syndrome
CSF	cerebrospinal fluid
CSH	carotid sinus hypersensitivity
CT	computerized tomography
CTX	cerebrotendinous xanthomatosis
DAT	dopamine transporter
DAVF	dural arteriovenous fistula
DBA	dopamine blocking agent
DBS	deep brain stimulation
DHPR-D	pyruvoyltetrahydropterin synthase deficiency
DIP	drug-induced parkinsonism
DLB	dementia with Lewy bodies
DMPS	2,3-dimercapto-1-propane-sulfonate
DMSA	dimercaptosuccinic acid
DRBA	dopamine receptor blocking agents
DRD	dopa-responsive dystonia
DRPLA	dentatorubropallidoluysian atrophy
DTI	diffusion tensor imaging
DWI	diffusion-weighted MRI imaging
EA1	episodic ataxia type 1
EA2	episodic ataxia type 2
EBV	Epstein-Barr virus
EDTA	edetic acid
EEG	electroencephalogram (or electroencephalography)
EL	encephalitis lethargica
EMG	electromyography (or electromyographic)
EMST	expiratory muscle strength training program
EPS	extrapyramidal side effects

Abbreviations

ERK	extracellular-signal-regulated kinase
ESRD	end-stage renal disease
ET	essential tremor
FAHN	fatty acid hydroxylase-associated neurodegeneration
FDG-PET	fluorodeoxy-glucose positron emission tomography
FHM	familial hemiplegic migraine
FHM1	familial hemiplegic migraine 1
FHM2	familial hemiplegic migraine 2
FHM3	familial hemiplegic migraine 3
FLAIR	fluid attenuated inversion recovery
FLE	frontal lobe epilepsy
GA1	glutaric aciduria type 1
GABA	gamma-aminobutyric acid
GAD	glutamic acid decarboxylase
GALT	galtactose-1-uridyltransferase
GAMT	guanidinoacetate methyltransferase
GBA	glucocerebrosidase gene
GCase	lysosomal hydrolase β-glucocerebrosidase
GCDH	glutaryl-CoA dehydrogenase
GD	Gaucher's disease
GEFS+	generalized epilepsy with febrile seizures plus
GH	growth hormone
GHS	Gordon Holmes spinocerebellar ataxia syndrome
GLUT-1	glucose transporter protein type 1
GnRH	gonadotrophin-releasing hormone
GOSR2	Golgi SNAP receptor complex 2 gene
GPe	globus pallidus (external segment)
GPi	globus pallidus (internal segment)
GSK3β	glycogen synthase kinase 3β
GTC	generalized tonic clonic
GTPCH1	guanosine triphosphate cyclohydrolase 1
GTS	Gilles de la Tourette syndrome
HAD	HIV/AIDS-associated dementia
HCHB	hemichorea-hemiballism
HCN	hyperpolarization-activated cyclic nucleotide-gated
HD	Huntington's disease
HH	hereditary hemochromatosis
HFE	human leukocyte antigen-H
HFO	high frequency oscillations
HFS	hemifacial spasm
HIV	human immunodeficiency virus
HKPP	hyperkalemic periodic paralysis
HL	Hodgkin's lymphoma
HLA	human leucocyte antigen
HMDPC	hypermanganesemia with dystonia, polycythemia, and cirrhosis
HOKPP	hypokalemic periodic paralysis
HOKPP1	hypokalemic periodic paralysis type 1
HOKPP2	hypokalemic periodic paralysis type 2
HOKPP3	hypokalemic periodic paralysis type 3
HP	*Helicobacter pylori*
HSV	herpes simplex virus
HVA	homovanillic acid
Hz	Hertz
IBS	irritable bowel syndrome
ICA	internal carotid artery
ICCA	infantile convulsions and paroxysmal choreoathetosis
IEG	immediate early gene
IEM	inborn error of metabolism
IFN	interferon
IGE	idiopathic generalized epilepsy
IgG	immunoglobulins
INAD	infantile neuroaxonal dystrophy
IPD	idiopathic Parkinson's disease
IRIS	immune reconstitution inflammatory syndrome
IRLSSG	International Restless Legs Syndrome Study Group
IV	intravenous
IVIg	intravenous immunoglobulin
JME	juvenile myoclonic epilepsy
JSRD	Joubert-syndrome-related disorder
Kcal	kilocalories
KD	ketogenic diet
LBD	Lewy body disease
LC	lethal catatonia
LDL	low-density lipoprotein
LGS	Lennox-Gastaut syndrome
LID	levodopa-induced dyskinesias
LIMP-2	lysosomal integral membrane protein type 2
LND	Lesch-Nyhan disease
LSA	limb shaking attacks
LSD	lysosomal storage disorders
LSVT	Lee Silverman Voice Treatment
MAO	monoamine oxidase
MAOI	monoamine oxidase inhibitor
MCM	methylmalonyl-CoA mutase
MD	movement disorder
MEG	magnetoencephalography
MELD	model for end-stage liver disease
MERRF	myoclonic epilepsy with ragged red fibers
MJD	Machado-Joseph disease
MLS	McLeod syndrome
MMA	methylmalonic aciduria
MND	mild neurocognitive disorder
MoS	Morvan's syndrome
MPAN	MIN-associated neurodegeneration
MPP+	1-methyl-4-phenylpyridinium
MPPP	1-methyl-4-phenyl-4-propionoxypiperidine
MPTP	1-methyl-4-phenyl-1,2,3,6-tetrahydropyridine
MRI	magnetic resonance imaging
MRS	magnetic resonance spectroscopy
MS	multiple sclerosis
MSA	multiple system atrophy
msec.	millisecond
MSN	medium spiny neurons
MSS	Marinesco-Sjögren syndrome
MST	multiple sclerosis tremor
MSUD	maple syrup urine disease

mTOR	mammalian target of rapamycin
MTP	microsomal triglyceride transfer protein
NAA	N-acetyl-aspartate
nACh	nicotinic acetylcholine
NBIA	neurodegeneration with brain iron accumulation
NCL	neuronal ceroid lipofuscinosis
NE	norepinephrine
NFLE	nocturnal frontal lobe epilepsy
NH	nocturnal hypertension
NHL	non-Hodgkin's lymphoma
NIC	neuroleptic-induced catatonia
NIID	neuronal intranuclear inclusion disease
NKH	non-ketotic hyperglycemia
NMDA	N-methyl-D-aspartate
NMDAR	NMDA glutamate receptor
NFLE	nocturnal frontal lobe epilepsy
NMS	neuroleptic-malignant syndrome
NP-C	Niemann-Pick type C
NPY	neuropeptide Y
NREM	non-rapid eye movement
NSAIDs	non-steroidal inflammatory drugs
OCD	obsessive-compulsive disorder
OH	orthostatic hypotension
OI	opportunistic infection
o.i.d.	once a day
OMS	opsoclonus-myoclonus syndrome
OSA	obstructive sleep apnea
PA	propionic acidemia (or aciduria)
PAF	pure autonomic failure
PANDAS	pediatric autoimmune neuropsychiatric disorders associated with *Streptococcus*
PANS	pediatric acute-onset neuropsychiatric syndrome
PA-S	periodic acid-Schiff
PAS	para-aminosalicylic acid
PaT	palatal tremor
PCD	paraneoplastic cerebellar degeneration
PCNSL	primary central nervous system lymphoma
PCR	polymerase chain reaction
PD	Parkinson's disease
PDC	parkinsonism-dementia complex of Guam
PDHc	pyruvate dehydrogenase complex
PDS	parkinsonism-dementia syndrome
PED	paroxysmal exertion- (or exercise-) induced dyskinesia
PEG	percutaneous endoscopic gastrostomy
PERM	progressive encephalitis with rigidity and myoclonus
PET	positron emission tomography
PHS	parkinsonism-hyperpyrexia syndrome
PIGD	postural instability gait disorder
PKA	protein kinase A
PKAN	pantothenate kinase-associated neurodegeneration
PKC	paroxysmal kinesigenic choreoathetosis
PKD	paroxysmal kinesiogenic dyskinesia
PLAN	PLA2G6-associated neurodegeneration
PLMD	periodic limb movement disorder
PLMS	periodic limb movements in sleep
PLP-DE	pyridoxal-phosphate-dependent epilepsy
PMC	paramyotonia congenital
PML	progressive multifocal leukoencephalopathy
PNKD	paroxysmal non-kinesigenic dyskinesia
POE	partial onset epilepsy
PPH	post-prandial hypotension
PSA	progressive system atrophy
PSG	polysomnography
PSP	progressive supranuclear palsy
PSP-RS	PSP-Richardson's syndrome phenotype
PTH	parathyroid hormone
PTPS-D	dihydropteridine reductase deficiency
PV	polycythemia vera
PVCM	paradoxical vocal cord motion
PVT	portal vein thrombosis
QSART	quantitative sudomotor axon reflex test
RBC	red blood cell
RBD	REM sleep behavior disorder
rCBF	regional cerebral blood flow
RDP	rapid onset dystonia-parkinsonism
RE	Rasmussen's encephalitis
REM	rapid eye movement
RF	rheumatic fever
RLS	restless legs syndrome
RMD	rhythmic movement disorders
RMMA	rhythmic masticatory muscle activity
RSSE	Russian spring-summer encephalitis
SAE	subcortical arteriosclerotic encephalopathy
SC	Sydenham's chorea
SCA	spinocerebellar ataxia
SCA6	spinocerebellar ataxia 6
SCD	sickle cell disease
SCLC	small-cell lung cancer
SEAT	steroid-responsive encephalopathy associated with autoimmune thyroiditis
SENDA	static encephalopathy of childhood with neurodegeneration in adulthood
SFG	spotted fever group
SH	supine hypertension
SIBO	small intestinal bacterial overgrowth
SjS	Sjögren's syndrome
SLE	systemic lupus erythematosus
SMEI	severe myoclonic epilepsy of infancy
SNpc	substantia nigra (pars compacta)
SNpr	substantia nigra (pars reticulate)
SPECT	single-photon emission computed tomography
SPS	stiff-person syndrome
SREAT	steroid-responsive encephalopathy associated with autoimmune thyroiditis
SS	superficial siderosis
SSEP	somatosensory evoked potential
SSPE	subacute sclerosing panencephalitis
SSRI	selective serotonin reuptake inhibitor
SuS	superficial siderosis
STN	subthalamic nucleus

Abbreviations

SV2A	synaptic vesicle glycoprotein 2A
TB	tuberculosis
TBE	tick-borne encephalitis
TbM	tuberculous meningitis
TB-RSSE	tick-borne Russian spring-summer encephalitis
TD	tardive dystonia
TH	tyrosine hydroxylase
THD	tyrosine hydroxylase deficiency
THMD2	thiamine metabolism dysfunction syndrome-2
TIBC	total iron binding capacity
t.i.d.	three times a day
TLE	temporal lobe epilepsy
TMP-SMX	trimethoprim-sulfamethoxazole
TMS	transcranial magnetic stimulation
TPO	thyroid peroxidase (or thyroperoxidase)
TS	Tourette's syndrome
TSH	thyroid-stimulating hormone
UAO	upper airway obstruction
ULS	Unverricht-Lundborg syndrome
UPDRS	Unified Parkinson's Disease Rating Scale
VGKC	voltage-gated potassium channel
VIM	ventral intermediate
VL	ventralis lateralis
VLDL	very-low-density lipoprotein
VOP	ventral oralis posterior
VP	ventral pallidum
VSD	ventral-septal defect
VZV	varicella-zoster virus
WD	Wilson's disease
WhD	Whipple's disease
XALD	X-linked adrenoleukodystrophy
ZI	zona incerta

Preface

Movement disorders are a group of neurological disorders often without an identifiable etiology or manifestations of genetic or neurodegenerative disorders. Classical examples include idiopathic Parkinson's disease, essential tremor, various forms of dystonia, chorea, myoclonus, tics, and other hypokinetic and hyperkinetic movement disorders. These and other movement disorders may also occur as presenting or prominent features of general neurological disorders such as stroke, encephalitis, multiple sclerosis, or CNS tumors. Moreover, they may also occur in the setting of a wide spectrum of systemic diseases, including metabolic and endocrine disorders, intoxications, hematological diseases, cancer, or infection. These latter situations where movement disorders are the hallmark of general neurologic or systemic conditions is when clinicians may face challenges of diagnosis and treatment. Affected patients may present to the internists and other physicians, including neurologists not skilled in movement disorders, who may be unfamiliar with the phenomenology or differential diagnosis.

We have invited an eminent group of international experts to contribute to this textbook, which we hope will serve as a reference guide to both neurologists and non-neurologists, residents, and consultant specialists who encounter patients with movement disorders in the setting of systemic or general neurological conditions. Following an introductory section summarizing the principles of the clinical approach to correctly classify movement disorder syndromes, their underlying pathophysiology, and their management, the different chapters provide comprehensive information on the clinical features, prevalence, pathophysiology, and diagnostic and management approach to specific classes of systemic or general neurologic conditions, where affected patients may present with movement disorders. We have also included a section addressing the important field of systemic complications occurring in the setting of primary movement disorder syndromes, including problems of swallowing, gastrointestinal dysfunction, orthostatic hypotension, and other types of autonomic failure, as well as movement disorder emergencies.

In addition to tables, diagrams, and illustrative figures of classical clinical or imaging findings, we have also included more than fifty video examples of typical movement disorders occurring in the various clinical settings discussed in this book. The videos, selected to further characterize the typical phenomenology and enhance clinical diagnostic skills, are available via the internet using QR technology.

Despite all our attempts to compile an authoritative and state-of-the-art textbook with chapters contributed from a faculty of outstanding international experts in their fields, this book will likely still leave room for improvement. We welcome constructive criticism from our readers and will use it to further refine, enlarge, and improve future editions. Meanwhile, we hope that this volume will be a useful and practical clinical companion to many junior as well as seasoned clinicians who encounter patients presenting with symptomatic movement disorders.

Werner Poewe
Joseph Jankovic
August 2013