OXFORD SPECIALIST HANDBOOKS IN PAEDIATRIC

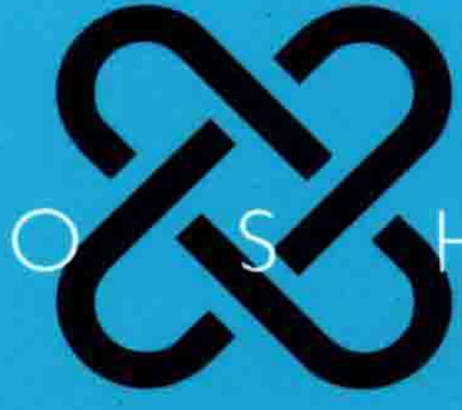
PAEDIATRIC NEUROLOGY

Edited by

Rob Forsyth

Richard Newton

SECOND EDITION



PAEDIATRICS

Oxford Specialist Handbooks in Paediatrics

Paediatric Neurology

Second edition

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Dedication

For our families:
Pip, Beth, and Ellen; Judith, Sarah, Michael, and Jennifer

And from all the contributors:
Thanks to our own families, and those we meet through our work, who support us and teach us so much.

Preface to the second edition

The very gratifying response to the first edition of this book has justified our belief in the value of a team approach: contributors with their fingers on the pulse of advances in our field, steadied at the helm by two editors with experience and perspective. It has, again, been a great privilege: never has Lord Acton's advice to 'learn as much from writing books as from reading them' been better heeded!

We have appreciated the very constructive suggestions for improvement of the first edition and followed them where we can. We have added respiratory consults to Chapter 5, and included more neuroradiology, diagrams, and images in situations where they offer clarity. A section on late-onset metabolic disease is added with an emphasis on how this group of disorders might catch us out.

With an ever-increasing list of genes and autoantibodies to think about it is important to remember those everyday skills we carry, honed as juniors in our specialty: listening to what is truly being said, careful clinical examination, focused investigation, and above all the communication of understanding, reassurance, and hope to families and young people facing challenges they never dreamed existed. You will find due emphasis on this in the text.

We trust this book will become a trusted companion. Please continue to tell us how it can be improved!

RF
RN
2012

Preface to the first edition

Medicine is a communal discipline, and this book has benefited greatly from being written in community by trainees (who remember the recent struggle to grasp a complex area) and older colleagues who can add particular emphases and perspective. We have striven to provide a combination of practical advice on clinical approach, and 'at a glance' oversights and *aides-memoire* to topic areas. We also wanted to address a number of practical issues that occupy a lot of time in practice, but that are rarely addressed in more conventional textbooks.

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2007

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2012

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Symbols and abbreviations

5FU	5-fluorouracil
5HIAA	5-hydroxyindoleacetic acid
5MTHF	5-methyl terahydrofolate
AAC	assistive/augmentative communication
AASA	L-alpha-aminoadipic semialdehyde
ABC	airway, breathing, circulation
ABG	arterial blood gases
ABI	acquired brain injury
ACE	angiotensin converting enzyme
AChE	anticholinesterase
AChR	acetylcholine receptor
ACTH	adrenocorticotrophic hormone
AD	autosomal dominant
ADANE	autosomal dominant acute necrotising encephalopathy
ADC	apparent diffusion coefficient (in MRI)
ADEM	acute disseminated encephalomyelopathy
ADHD	attention deficit hyperactivity disorder
ADP	adenosine diphosphate
ADI/ADOS	Autism Diagnostic Inventory/Observation Schedule
A&E	Accident and Emergency
AED	anti-epileptic drug
AFB	acid-fast bacilli
AFP	alpha-foeto protein
AGS	Aicardi–Goutièrres syndrome
AIDP	acute inflammatory demyelinating neuropathy
AIDS	acquired immunodeficiency syndrome
ALL	acute lymphoblastic leukaemia
ALT	alanine amino-transaminase
ALTE	acute life-threatening events
AMAN	acute motor axonal neuropathy
AMC	arthrogryposis multiplex congenital
AMP	adenosine monophosphate
AMPA(-R)	α-amino-3-hydroxy-5-methyl-4-isoxazole proprionic acid (receptor)
AMSAN	acute motor and sensory axonal neuropathy
ANA	antinuclear antibody

ANCA	antineutrophil cytoplasmic antibodies
AOA	ataxia oculomotor ataxia
AP	anteroposterior
APD	afferent pupillary defect
AR	autosomal recessive
ART	anti-retroviral treatment
ASOT	anti-streptolysin O titre
AST	aspartate aminotransferase
AT	ataxia telangiectasia
ATM	ataxia telangiectasia mutated protein
ATP	adenosine tri-phosphate
AV	arterioventicular
AVED	ataxia with vitamin E deficiency
AVM	arteriovenous malformation
BAER/P	brainstem auditory evoked response/potential
BAS	British Ability Scales
BBB	blood–brain barrier
BCNU	bis-chloroethylnitrosourea or carmustine
BCG	Bacillus Calmette–Guérin
bd	twice daily
BECTS	benign epilepsy with centrotemporal spikes
BFNS	benign familail neonatal seziures
BIND	bilirubin-induced neurological dysfunction
BIPAP	bilevel positive airway pressure
BM	(inf.) glucose monitoring strip
BMEI	benign myoclonic epilepsy of infancy
BMD	Becker muscular dystrophy
BMT	bone marrow transplant
BNF-C	British National Formulary for Children
BNS	benign neonatal syndrome
BP	blood pressure
BSD	brainstem death
CACH	childhood ataxia with CNS hypomyelination (Vanishing White Matter disease)
CADASIL	cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
CAE	childhood absence epilepsy
CAMHS	child and adolescent mental health services
CBF	cerebral blood flow
CBT	cognitive-behavioural therapy
CBZ	carbamazepine

CD	controlled drug
CDG	congenital disorder of glycosylation (also known as carbohydrate deficient glycoprotein syndrome)
CEOP	childhood epilepsy with occipital paroxysms
CFAM	cerebral function analysing monitor
CFEOM	congenital fibrosis of extraocular muscles
CFS/ME	chronic fatigue syndrome/myalgic encephalomyelopathy
CGD	chronic granulomatous disease
CGH	comparative genome hybridisation
CHARGE	coloboma, heart defects, (choanal) atresia,(growth) retardation, genital/urinary and ear abnormalities (syndrome)
CHAT	checklist for Autism in Toddlers
CIDP	chronic inflammatory demyelinating neuropathy
CINCA	chronic infantile neurological cutaneous and articular (syndrome)
CIS	clinically isolatated syndrome
CJD	Creutzfeld-Jakob disease
CK	creatinine (phospho)kinase
CKMB	muscle–brain isoenzyme of CK
CMAP	compound muscle action potential
CMD	congenital muscular dystrophy
CMS	congenital myaesthenic syndrome
CRMCC	cerebro-retinal microangiopathy with calcification and cysts
CMT	Charcot-Marie tooth Disease
CMV	cytomegalovirus
CNS	central nervous system
CNV	copy number variant
COACH	cerebellar vermis hypo- or aplasia, oligophrenia, congenital ataxia, ocular coloboma and hepatic fibrosis
COX	cytochrome oxidase
CPEO	chronic progressive external ophthalmoplegia
CP	cerebral palsy
CPP	cerebral perfusion pressure
C/R	controlled-release
CRMCC	cerebroretinal microangiopathy with calcification and cysts
CRP	C-reactive protein
CRPS	complex regional pain syndrome
CSE	convulsive status epilepticus
CSF	cerebrospinal fluid

CSI	craniospinal irradiation
CSW	cerebral salt wasting
CSWS	continuous spike-wave discharges during slow wave sleep
CT	computed tomography
CTA	computed tomographic angiography
CTG	cardiotocograph
CVA	cerebrovascular accident
CVI	cerebral (or cortical) visual impairment
CVID	common variable immunodeficiency
CVP	central venous pressure
CVVH	continuous veno-venous haemofiltration
CXR	chest X-ray
DA	dopamine
DAMP	disorders of attention, motor processing, and perception
DCD	developmental co-ordination disorder
DDAVP	desmopressin
DEXA	dual-energy X-ray absorptiometry
DI	diabetes insipidus
DIDMOAD	diabetes insipidus, diabetes mellitus, optic atrophy and deafness.
DIPG	diffuse intrinsic brainstem gliomas
DKA	diabetes ketoacidosis
DM	myotonic dystrophy
DMD	Duchenne muscular dystrophy
DML	distal motor latency
DNET	dysembryoplastic neuroepithelial tumour
DRD	L-DOPA responsive dystonia
DRPLA	dentato-rubral-pallido-luysian atrophy
DSA	digital subtraction angiography
DSM-IV	diagnostic and statistical manual of mental disorders–Fourth edition
DTI	diffusion tensor imaging (MRI)
DTR	deep tendon reflex
DWI	diffusion-weighted image (MRI)
EBV	Epstein–Barr virus
ECG	electrocardiogram
ECMO	extra-corporeal membrane oxygenation
EDH	extra-dural haemorrhage
EEG	electroencephalography
EIEE	early infantile epileptic encephalopahty (Ohtahara syndrome)

ELISA	enzyme-linked immunosorbent assay
EM	electron microscopy
EMA	epileptic absence syndrome
EMAS	epilepsy with myoclonic-astatic seizures
EMD	Emery–Dreyfuss muscular dystrophy
EME	early myoclonic encephalopathy
EMG	electromyography
ENT	ear, nose, and throat
EOG	electrooculogram
EOS	early onset sarcoid
EPC	epilepsia partialis continua
EPI	epinephrine
ERG	electroretinogram
ESES	electrical status during slow-wave sleep (synonymous with CSWS)
ESR	erythrocyte sedimentation rate
ET	endotracheal tube
EVD	extra-ventricular drain
FA	Friedreich ataxia
FAC	fatty acid
FEES	functional endoscopic evaluation of swallowing
FII	factitious or induced illness
FBC	full blood count
FCMD	Fukuyama congenital muscular dystrophy
FDG-PET	flurodeoxy-glucose positron emission tomography
FIM	functional independence measure
FISH	fluorescent in-situ hybridisation
FLAIR	fluid attenuated inversion recovery–MRI sequence
fMRI	functional MRI
FRAX	fragile X–locus
FSH	facioscapulohumeral muscular dystrophy
FTA-ABS	fluorescence trepoanema antibody-absorbed syndrome
FTT	failure to thrive
FVC	forced vital capacity
GA	general anaesthesia
GAA	guanidinoacetate
GABA	gamma amino-butyric acid
GAG	glycosaminoglycans
GAMT	guanidinoacetate methyltransferase
GBS	Guillain-Barré syndrome
GC-MS	gas chromatography mass spectroscopy

GCS	Glasgow coma score
GCT	germ cell tumour
GEFS+	generalized epilepsy with febrile seizures plus
GHB	gamma hydroxy-butyrate
GI	gastrointestinal
GIT	gastrointestinal tract
GLUT1 DS	glucose transporter enzyme 1 deficiency syndrome
GM	ganglioside
GM-CSF	granulocyte–macrophage colony-stimulating factor
GMFCS	gross motor function classification syndrome
GMFM	gross motor function measure
GMH	germinal matrix haemorrhage
GMPM	gross motor performance measure
GORD	gastro-esophageal reflux disease
GSD	glycogen storage disease
GTC	generalized tonic-clonic (seizure)
HAART	highly active anti-retroviral therapy
HCG	human chorionic gonadotrophin
HDU	high dependency unit
HELLP	haemolysis, elevated liver enzymes with low platelet count
HHV	Herpes hominis virus
HIE	hypoxic-ischaemic encephalopathy
HIV	human immunodeficiency virus
HLH	haemophagocytic lymphohistiocytosis
HLA	human leucocyte antigen
HMN	hereditary motor neuropathy
HMSN	hereditary sensory motor neuropathy
HNPP	hereditary neuropathy with liability to pressure palsies
HOCUM	hypertrophic obstructive cardiomyopathy
HPE	HIV-associated progressive encephalopathy
HSAN	hereditary sensory and autonomic neuropathy
HSV	Herpes simplex virus
HUS	haemolytic uraemic syndrome
HVA	homo-vanilic acid
IBD	inflammatory bowel disease
ICD-10	International Classification of Diseases v.10
ICF	International classification of Functioning, Disability and Health
ICP	intracranial pressure
ICU	intensive care unit

ID	infectious diseases
IDDM	insulin dependent diabetes mellitus
IEF	iso-electric focusing
IEM	inborn errors of metabolism
IFNα	interferon-α
IIH	idiopathic intracranial hypertension
ILAE	International League against Epilepsy
IM	intramuscular
INAD	infantile neuraxonal dystrophy
INO	internuclear ophthalmoplegia
INR	international normalization ratio
IOP	intraocular pressure
IPH	intraparenchymal haemorrhage
IS	infantile spasms
IT	intrathecal
IUGR	intrauterine growth retardation
IV	intravenous
IVA	isovaleric acidaemia
IVH	intraventricular hemorrhage
IVIG	intravenous immunoglobulin
JAE	juvenile absence epilepsy
JEV	Japanese encephalitis virus
JIA	juvenile idiopathic arthritis
JME	juvenile myoclonic epilepsy
JRA	juvenile rheumatoid arthritis
KD	Kawasaki disease
KF	Kaiser-Fleischer (ring)
KSS	Kearn–Sayre syndrome
LCMV	lymphocytic choriomeningitis virus
L-DOPA	L-3,4-dihydroxyphenylalanine (levodopa)
LD	learning difficulty
LDH	lactate dehydrogenase
LE	lupus erythematosus (also a particular cell seen in blood film in lupus erythematosus
LEV	levetiracetam
LFT	liver function test
LGMD	limb girdle muscular dystrophy
LGS	Lennox-Gastaut syndrome
LHON	Leber's hereditary optic neuropathy
LKS	Landau-Kleffner syndrome

LMN	lower motor neurone
LMP	last menstrual period
LMWH	low molecular weight heparin
LP	lumbar puncture
MABC	movement assessment battery for children
MAO	mono-amine oxidase
MAP	mean arterial pressure
MCA	middle cerebral artery
MCAD	medium chain acyl coenzyme A
MC&S	microscopy, culture, and sensitivity
MCT	medium chain triglyceride
MD	muscular dystrophy
MEB	muscle eye brain disease
MECP2	methyl-CpG-binding protein 2 gene–common Rett syndrome gene
MELAS	mitochondrial encephalomyopathy, lactic acidosis, and strokelike episodes
MERRF	myoclonic epilepsy and ragged-red fibre
MGUS	monoclonal gammopathy of unknown significance
MH	malignant hyperthermia
MIBG	meta-iodobenzyl guanidine (Iodine123)
MLD	metachromatic leucodystrophy
MLF	medial longitudinal fasciculus
MLPA	multiplex ligation-dependent probe amplification
MLST	multiple sleep latency test
MMA	methylmalonic acidaemia
MMF	mycophenylate mofetil
MMR	mumps, measles and rubella
MMSE	mini mental state examination
MNGIE	myopathy and external ophthalmoplegia; Neuropathy; Gastro-Intestinal; Encephalopathy
MPGR	multi-planar gradient echo sequence–on MRI
MPS	mucopolysaccharidosis
MRA	magetic Resonance Angiography
MRC	Medical Research Council
MRI	magnetic resonance imaging
MRS	magnetic resonance spectroscopy
MRV	magnetic resonance venography
MS	multiple sclerosis
MS/MS	tandem mass spectroscopy
MSUD	maple syrup urine disease

MTHFR	methylene tetrahydrofolate
MUP	motor unit potential
MuSK	musculo-specific kinase
MWT	maintenance of wakefulness test
NADH	nicotinamide adenine dinucleotide
NADH-TR	NADH-tetrazolium reductase
NAI	non-accidental injury
NARP	neuropathy, ataxia and retinitis pigmentosa
NBIA	neurodegeneration with brain iron accumulation
NCC	neurocysticercosis
NCL	neuronal ceroidal lipofuscinosis
NCSE	non-convulsive status epilepticus
NCV	nerve conduction velocity
NE	norepinephrine
NEC	necrotizing enterocolitis
NEAD	non-epileptic attack disorder
NF	neurofibromatosis
NGT	nasogastric tube
NICE	National Institute of Clinical Excellence
NICU	neonatal intensive care unit
NIH	National Institutes of Health
NIPPV	non-invasive positive pressure ventilation
NKH	non-ketotic hyperglycinemia
NMD	neuromuscular disease
NMDA(-R)	N-methyl-D-aspartate (receptor)
NMO	neuromyelitis optica
NPV	negative predictive value
NSAID	non-steroidal anti-inflammatory
NTD	neural tube defect
OCD	obsessive compulsive disorder
OCP	oral contraceptive pill
od	once daily
OFC	occipitofrontal circumerence
OGB	oligoclonal bands
OGD	oesophago-gastro-duodenoscopy
OKN	optokinetic nystagmus
OMS	opsoclonus-myoclonus syndrome (Kinsbourne)
ON	optic neuritis
OSA	obstructive sleep apnoea
OT	occupational therapist