

Cambridge University Press & Assessment 978-1-108-74418-8 — Neuromuscular Disease 2nd Edition Jessica E. Hoogendijk , Marianne de Visser , Pieter A. van Doorn , Erik H. Niks Index <u>More Information</u>

Index

Locators in bold refer to tables; those in italic to figures; underline to videos

```
acetylcholine receptor (AChR)
  congenital myasthenic syndromes,
     170
  neuromuscular junction disorders,
AChR-myasthenia gravis, 157-158, 160
  clinical features, 157
  clinical history/symptoms, 157, 158
  compound muscle action potential,
  diagnosis, 157, 158
  examination, 157
  follow-up, 157
  management, 158-161
  myasthenic crisis, 161
  prognosis, 158-161
acid alpha-glucosidase (GAA), 29-30,
     219
acid maltase see acid alpha-glucosidase
acute flaccid myelitis (AFM), 93
acute inflammatory demyelinating
     polyneuropathy (AIDP), 96
acute motor and sensory axonal
     neuropathy (AMSAN), 96
acute motor axonal neuropathy
    (AMAN), 96
adrenomyeloneuropathy, 78
AFM (acute flaccid myelitis), 93
AIDP (acute inflammatory
     demyelinating polyneuropathy), 96
alcohol-induced myopathies, 263, 264,
     265
alcoholic polyneuropathy, 130-131, 138
  clinical history/symptoms, 130
  diagnosis, 130
  examination, 130
  follow-up, 130
  management, 131
  prognosis, 131
aldolase A deficiency (GSDX11,
    ALDOA), 33
allodynia, 28, 110, 132
alpha-dystroglycan, 191-192
ALS see amyotrophic lateral sclerosis
AMAN (acute motor axonal
     neuropathy), 96
amiodarone polyneuropathy, 137-139
  blue-grey discoloration of face/hands,
     138
  clinical history/symptoms, 137
  diagnosis, 137
  examination, 137
  follow-up, 137
AMSAN (acute motor and sensory
```

```
clinical history/symptoms, 1, 71
  diagnosis, 1-2, 71-72, 73
  differential diagnoses, 74-75
  electrodiagnostic studies, 37
  examination, 71
  follow-up, 72
  Gold Coast criteria for diagnosis, 74
  management, 75-76
  multidisciplinary care, 76
  phenotypes, 72
  spasticity management, 60
  videos, 272
anaesthetic management, 60-63
Andersen-Tawil syndrome, 213, 215
ANO5 distal myopathy, 23-24
ANO5 gene, 202
anoctaminopathy, 203-205, 204
anterior horn cell diseases, 1-2
antisense oligonucleotide mediated exon
     skipping, 174
antisense oligonucleotides (ASOs), 57
anti-synthetase syndrome (ASyS), 45,
     249
arachnodactyly, MTM1 myopathy, 238,
     239
ASOs see antisense oligonucleotides
AsyS see anti-synthetase syndrome
ataxia, differential diagnoses, 26
ATTR see transthyretin amyloidosis
ATTR-PN (transthyretin amyloidosis-
     polyneuropathy), 155, 156
autonomic dysfunction, examination, 14
axial weakness
  differential diagnoses, 19-20
  examination, 10
azathioprine, 160, 162, 248, 255
Becker muscular dystrophy (BMD), 174,
     177 - 179
  clinical history/symptoms, 176, 176
  CT scan, 177
  diagnosis, 176
  differential diagnoses, 176
  examination, 176
  follow-up, 176
  imaging, 45
  incidence, 177
  muscle biopsy, 177, 179
  phenotypical spectrum of
     dystrinopathies, 178
  videos, 273
Becker myotonia, 213, 215
behavioural problems, 7-8, 174
Beighton scale for hypermobility, 9
bent spine (camptocormia), 19-20
```

amyotrophic lateral sclerosis (ALS), 1,

72-76, 73

```
β enolase deficiency, 33
Bethlem myopathy, 193-194
  clinical features, 194
  clinical history/symptoms, 193
  CT scan, 195
  diagnosis, 193
  elbow contractures, 193
  examination, 193
  imaging, 45
biopsy see muscle biopsy; nerve biopsy;
    skin biopsy
blue-grey skin discoloration,
    amiodarone polyneuropathy, 137,
BMD see Becker muscular dystrophy
Borrelia burgdorferi see Lyme
    radiculopathy/Lyme disease
bortezomib, 107, 138
botulism, 29-30, 86
brachial plexus neuropathy see neuralgic
    amyotrophy
breathing problems see respiratory
    muscle weakness
Brody disease, 9, 32-34, 228
Brown-Violetta-Van Laere syndrome,
    16, 18
bulbar muscles; see also spinal and
    bulbar muscular atrophy
  DM1, 184
  examination, 9-10
  MG, 17
  postpolio syndrome, 92
CACNA1S gene, 213
  exertional rhabdomyolysis, 269, 269
calpain-related limb girdle muscular
     dystrophy, 189-190, 190
calpainopathy, 189-190, 190, 273
camptocormia (bent spine), 19-20
cannabis-derived products, neuropathic
     pain, 60
CANOMAD syndrome (chronic ataxic
     neuropathy, external
     ophthalmoplegia, M-protein
    agglutination, disialosyl
    antibodies), 16
carbohydrate metabolic myopathies,
    224; see also McArdle disease;
     Pompe disease
cardiac function, 30-31
  alcohol use, 265
  amyloidosis, 155, 156
  Andersen-Tawil syndrome, 213, 215
  BMD, 178, 179
  desminopathy, 211
  diagnosis, 6
  DM, 249
```

axonal neuropathy), 96

amyloidosis, 155, 155

amyloid myopathy, 49, 249-250, 252



Cambridge University Press & Assessment 978-1-108-74418-8 — Neuromuscular Disease 2nd Edition

Jessica E. Hoogendijk, Marianne de Visser, Pieter A. van Doorn, Erik H. Niks

More Information

Index

DM1, 184 congenital myasthenic syndromes chronic idiopathic axonal DM2, 185, 186, 186 polyneuropathy (CIAP), 134 (CMS), 3, 170; see also Dok7 drug-induced myopathies, 263-264 clinical features, 134 drug-induced polyneuropathies, 137 diagnosis, 171 clinical history/symptoms, 131-132 EDMD, 199, 199-200 diagnosis, 132 management, 59 differential diagnoses, 133 MFMs, 210, 211 examination, 132 videos, <u>273</u> myopathies, 4, 208 follow-up, 132 OPMD, 197 risk factors, 134 PMDs, 232 chronic inflammatory demyelinating Pompe disease, 221 polyneuropathy (CIDP), 108 pregnancy, 63 clinical history/symptoms, 97 carnitine palmitoyltransferase (CPT) diagnosis, 97-98 deficiency, 33-34 diagnostic criteria, 97 clinical history/symptoms, 227 differential diagnoses, 43, 98 diagnosis, 227-228 distribution of symptoms in variants, corticosteroids follow-up, 228 types, 228-229 examination, 97 cataract, 6-7, 32-34, 53, 185 follow-up, 98 CAV3 gene, 201 imaging, 42-43, 44 counselling, 65 caveolinopathy, 123, 201 management, 98-99 FSHD, 182 clinical features, 201 prognosis, 98-99 OPMD, 197 clinical history/symptoms, 200 videos, 273 diagnosis, 201 chronic polyneuropathy, 132-134; see examination, 200-201 also above follow-up, 201 differential diagnoses, 133 videos, 273 prevalence, 133 cramp, 178 central core disease, 19-20, 32, 235, 236, risk factors, 134 236, 236 chronic progressive external examination, 8 central nervous system involvement, ophthalmoplegia (CPEO), 230, management, 60 7-8, 10, **22-23**, 123, **231**, 234-235 232, 233 cerebrospinal fluid, 1-2, 93, 94 clinical history/symptoms, 230, 233 cervical spondylotic myelopathy, 74-75 clinical features, 232 cervical stenosis, 78, 152 differential diagnoses, 230 channelopathies see skeletal muscle BMD, 179 examination, 230 channelopathies follow-up, 230 IIMs, 249 Charcot arthropathy, 153 management, 233 JDM, 244 Charcot deformities, 151 CIAP see chronic idiopathic axonal Charcot-Marie-Tooth disease (CMT), polyneuropathy 64, 273 CIDP see chronic inflammatory Charcot-Marie-Tooth disease type 1A, demyelinating polyneuropathy 144-148 CIP/CIM see critical illness classificatory features, 145-146 polyneuropathy/myopathy clinical features, 146 CIPN see chemotherapy-induced clinical history/symptoms, 144 peripheral neuropathy diagnosis, 144 cisplatin, 24-25, 26 diagnosis, 135 differential diagnoses, 145-146 CK see creatine kinase examination, 144 claw hand follow-up, 136 follow-up, 144 CMT, 144, 144, 145-146 management, 147 leprosy, 142, 142 muscle atrophy, 147 CMS see congenital myasthenic Charcot-Marie-Tooth disease type 2, syndromes 145-146, 145-146 CMT see Charcot-Marie-Tooth disease BMD, 177 Charcot-Marie-Tooth disease (CMT) LGMD, 188, 191 CNV analysis see copy number variant types 2A/2B, 150-151 analysis MG, 157 clinical history/symptoms, 149, 149 coasting effect, 24-25, 139 diagnosis, 150 Coats syndrome, 181 examination, 149-150 coenzyme Q 10, 33 - 34follow-up, 150 collagen VI-related myopathies; see also RAS-associated protein gene, 150, Bethlem myopathy; Ullrich 150-151 congenital muscular dystrophy chemotherapy-induced peripheral clinical features, 194 neuropathy (CIPN), 138, 138 imaging, 45 complex repetitive discharges (CRDs), chloride channelopathy, 212 chloroquine (CQ), drug-induced EMG, 272 dapsone, 138, 143 myopathies, 263-264 congenital cranial dysinnervation DCM (dilated cardiomyopathy), 178, CHRNE gene, 170 disorders (CCDDs), 16 179 chronic fatigue syndrome, congenital fibrosis of the extraocular deafness, 16, 145-146

muscles (CFEOM), 16

myasthenic syndrome genes involved, 171 management, 171-172 congenital myopathies, imaging, 45; see also nemaline myopathy congenital ptosis, 16 contractures see elbow contractures; muscle contractures copper deficiency myelopathy, 78 copy number variant (CNV) analysis, 54; see also genetic testing drug-induced myopathies, 264, 265 immunotherapy, 57-58 vasculitic neuropathy, 109-110 spinal muscular atrophy, 86 CPEO see chronic progressive external ophthalmoplegia CPT 2, see carnitine palmitoyltransferase differential diagnoses, 27 cramp-fasciculation syndrome, 123, 124 creatine kinase (CK), 268; see also hyperCKaemia Bethlem myopathy, 194 McArdle disease, 224 myopathies, 40-41 non-dystrophic myotonic syndromes, critical illness polyneuropathy (CIP)/ myopathy (CIM), 136 clinical features, 136 clinical history/symptoms, 135 examination, 135 ICU-acquired weakness, 136 ventilator weaning, 136-137 CT scanning (computed tomography) Bethlem myopathy, 195 cyanocobalamin deficiency, 78 cytochrome b deficiency (Complex III), cytochrome-C-oxidase deficiency, cytostatic medication, drug-induced polyneuropathies, 138, 138 Danon disease, 30-31, 32

deep finger flexor muscles, 74-75

165-166



Cambridge University Press & Assessment 978-1-108-74418-8 — Neuromuscular Disease 2nd Edition Jessica E. Hoogendijk , Marianne de Visser , Pieter A. van Doorn , Erik H. Niks **More Information**

Index

Dejerine-Sottas phenotype, 146	clinical features, 170-171	myopathies, 40–41
dementia, frontotemporal	clinical history/symptoms, 168–169	neuromuscular junction disorders,
ALS, 7, 53, 73	diagnosis, 169	38, 40
VCP distal myopathy, 19–20 , 208	examination, 169	neuropathies, 35, 38, 39
	follow-up, 169–170	PPS, 92
demyelination	management, 171–172	videos, 271–272
CMT type 2, 145–146	muscle biopsy, 169	emerinopathies, imaging, 45
drug-induced polyneuropathies, 137,	muscle ultrasound, 170	emerins (nuclear envelope proteins), 200
138	dorsal root ganglia (DRG), 114–115, 115	Emery–Dreifuss muscular dystrophy
GBS, 96	dropped head, differential diagnoses,	(EDMD), 199–200
neuropathies, 38	19–20	clinical features, 199
dermatomyositis (DM), 251	drug-induced myasthenia gravis	
clinical features, 249		clinical history/symptoms, 198
clinical history/symptoms, 247, 247,	clinical history/symptoms, 163	diagnosis, 198–199
250, 251	diagnosis, 164	examination, 198
dermatomyositis-specific	drugs to avoid or use with caution, 165	follow-up, 199
autoantibodies, 250		MRI scan, 199
diagnosis, 247	examination, 163–164	phenotypes, 200
differential diagnoses, 249–250	follow-up, 164	EMG (electromyography) see
examination, 247	management, 165	electrodiagnostic studies
follow-up, 247–248	drug-induced myopathies, 263, 264 , 265;	end-of-life care see palliative care
imaging, 45	see also hydroxychloroquine	endocrine myopathy, 261–262
muscle biopsy, 249–250	myopathy	clinical history/symptoms, 260
short tau inversion recovery data, 248	alcohol, 263, 264 , 265	diagnosis, 260–261
skin abnormalities, 250	clinical history/symptoms, 262	differential diagnoses, 260
dermatomyositis-specific autoantibodies	diagnosis, 263	examination, 260
(DMSAs), 250	examination, 263	follow-up, 261
DES gene, 209	follow-up, 263	muscle biopsy, 261
desminopathy, 211	hydroxychloroquine, 263–264, 264	eosinophilic granulomatosis with
diabetic neuropathy, 128-129	muscle biopsy, 263	polyangitis (EGPA), 109
classificatory features, 129	statins, 263, 264 , 264–265	epimerase-kinase enzyme, 207; see also
clinical history/symptoms, 127	steroids, 264 , 265	GNE myopathy
diagnosis, 127-128	vitamin D deficiency, 263, 264 , 266	Erasmus Polyneuropathy Symptom
examination, 127	drug-induced polyneuropathies,	Score (E-PSS), 132
follow-up, 128	137–139	ERM see exertional rhabdomyolysis
management, 129	clinical history/symptoms, 137	EURO-NMD-ERN recommendations,
nerve injury patterns, 128	cytostatic drugs, 138, 138	immunohistochemical stains, 49
diaphragm weakness, 87, 126, 137,	diagnosis, 137	European Federation of Neurological
145–146, 164, 192	examination, 137	Societies/Peripheral Nerve Society
dilated cardiomyopathy (DCM), 178,	toxins and drugs which can cause, 138	(EFNS/PNS), MMN diagnostic
179	Duane syndrome, 16	criteria, 119
distal anoctaminopathy, 202-203	Duchenne muscular dystrophy (DMD),	European Malignant Hyperthermia
distal filaminopathy, 204	174–175	(MH) Group, 237
distal myopathies, 22-23, 203, 205; see	clinical history/symptoms, 173, 173	exertional rhabdomyolysis (ERM)
also GNE myopathy	comorbidities, 174	clinical history/symptoms, 269, 269
clinical history/symptoms, 202, 202,	diagnosis, 173	diagnosis, 269–270
204	DMD gene variants, 174, 175	examination, 269
diagnosis, 202-203	examination, 173	follow-up, 270
differential diagnoses, 23-24,	follow-up, 173-174	RYR1 myopathy, 237
202-203	imaging, 45	external ophthalmoplegia, 9, 16
examination, 202	phenotypical spectrum of	
follow-up, 203	dystrinopathies, 178	facial weakness
short tau inversion recovery data, 203	dysautonomia, differential diagnoses, 26	differential diagnoses, 17
showing rimmed vacuoles in muscle	DYSF gene, 202, 203	examination, 9
biopsy, 208	dysferlinopathy, 203, 204, 205	facioscapulohumeral muscular
distal myopathy with myotilin defect,	dysimmune neuropathies, 26–27, 38, 43,	dystrophy (FSHD), 181–182
204	272	clinical features, 181
distal myopathy with rimmed vacuoles	dysphagia, management, 59-60	clinical history/symptoms, 180
see GNE myopathy	dystroglycanopathies, 191-192	diagnosis, 180
DM see myotonic dystrophy	dystrophin-glycoprotein complex	examination, 180
DMD gene variants, 174, 175, 178 ; see	(DGC), 190	follow-up, 180
also Becker muscular dystrophy;		imaging, 45
Duchenne muscular dystrophy	EDMD see Emery-Dreifuss muscular	phenotypes, 181
DMRV (distal myopathy with rimmed	dystrophy	pregnancy and obstetrics, 64
vacuoles) see GNE myopathy	elbow contractures, Bethlem myopathy,	family history
DMSAs (dermatomyositis-specific	193	genetic testing, 53
autoantibodies), 250	electrodiagnostic studies, 35, 36, 39	history taking, 6–7
DNA testing see genetic testing	ALS, 37	fasciculations
DNAJB6 gene, 21–22, 23–24	IgM anti-MAG polyneuropathy, 103	differential diagnoses, 27
Dok7 myasthenic syndrome, 170	motor neuron diseases, 35	examination, 8

276

Dok7 myasthenic syndrome, 170



Cambridge University Press & Assessment 978-1-108-74418-8 — Neuromuscular Disease 2nd Edition

Jessica E. Hoogendijk , Marianne de Visser , Pieter A. van Doorn , Erik H. Niks

Index

More Information

Index

diagnosis, 267

examination, 267

follow-up, 267

MRI scan, 267

examination, 9

differential diagnoses, 32, 267

drug-induced myopathies, 264

hyperkalaemic periodic paralysis, 213

hypermobility/hyperlaxity
Beighton scale for hypermobility, 9

differential diagnoses, 28-29

hyperthyroid myopathy, 262

ultrasound, 272 fatigability/fatigue examination, 11-13 management, 58 SMA type 3, 90 tests, 13 fatty acid metabolism disorders, 33-34 fatty acid oxidation disorders (FAODs), 228, 229 Fazio-Londe syndrome, 18 fibreoptic evaluation of swallowing (FEES), 10 FKRP gene see limb girdle muscular dystrophy R9 flail arm syndrome, 72, 81, 83 forced vital capacity, 10, 71, 72, 80, 96, 164, 207, 211 Friedreich ataxia, 28-29 frontotemporal dementia see dementia FSHD see facioscapulohumeral muscular dystrophy functional neurological symptom disorder (FND), 14 GAA (acid alpha-glucosidase), 219 ganglionopathies, 114-115; see also paraneoplastic sensory neuronopathy gastrointestinal involvement, management, 59-60 GBS see Guillain-Barré syndrome gene-phenotype associations, 55 genetic(s) differential diagnoses, 146 distal myopathies, 203 DM2, 185-186 EDMD, 200 FSHD, 181 MTM1 myopathy, 240 myofibrillar myopathies, 210-211 OPMD, 196-197 peripheral neuropathies, 2 RYR1 myopathy, 237 SMA type 1, 86, 87 SMA type 3, 88-90 genetic counselling see counselling genetic testing, 52, 52 consequences of genetic diagnosis, 53 family history, 53 first-line tests, 53-54 interpretation of results, 55-56 mitochondrial DNA, 54 NGS panels, 53-54 second-line tests, 54 structural variations, 54 genetic treatments, 57 genetic variants, significance of, 55-56 genome aggregation database (gnomAD), 55 Gestalt approaches, ix, 47 glutaric aciduria type I, 33-34 glycogen metabolism disorders, 33 glycogen storage diseases see McArdle disease; Pompe disease GNE myopathy, 207-209 clinical features, 208 clinical history/symptoms, 206, 207 diagnosis, 206-207

follow-up, 207 MRI scan, 207 pregnancy and obstetrics, 64 rimmed vacuoles in muscle biopsy, 207-208, 208 Gottron papules, DM, 247, 251 Gowers sign DMD, 173 videos, 273 Graves disease, 213, 214, 262 GSDs (glycogen storage diseases) see McArdle disease; Pompe disease Guillain-Barré syndrome (GBS), 94-96 clinical history/symptoms, 94 diagnosis, 94 diagnostic criteria, 95 examination, 94 follow-up, 94 frequency of disease, 95 imaging, 43 immunotherapy, 57 management, 96 MFS-GBS overlap syndrome, 96, 272 prognosis, 96

Hashimoto thyroiditis, 261, 262 heart see cardiac function hereditary inclusion body myopathy see GNE myopathy hereditary motor neuropathies (HMN), diagnosis, 1-2 therapies, 2 hereditary neuralgic amyotrophy (HNA), 126-127 hereditary neuropathy with liability to pressure palsies (HNPP), 145-146, 147-148 hereditary sensory and autonomic neuropathies (HSAN), 145-146, hereditary sensory and autonomic neuropathy type 4, 150, 152-153, Charcot arthropathy, 153 clinical features, 152 clinical history/symptoms, 151, 151 diagnosis, 151-152 examination, 151 follow-up, 152 MRI scan, 152 hereditary spastic paraplegia (HSP), **28–29**, **78**, 153 hereditary transthyretin amyloidosispolyneuropathy (ATTRv-PN), 155, 156; see also transthyretin amyloidosis Hirayama disease, 22-23, 74-75, 83 histochemical stains, muscle biopsy, 49 HMN see hereditary motor neuropathies HNA (hereditary neuralgic

amyotrophy), 126-127

HSAN see hereditary sensory and

autonomic neuropathies

hydroxychloroquine, drug-induced

myopathies, 263-264, 264

clinical history/symptoms, 266-267

hyperCKaemia (elevated creatine

kinase), 268

hypokalaemic periodic paralysis (HypoPP), 213 clinical history/symptoms, 216 diagnosis, 216 examination, 216 follow-up, 217 short tau inversion recovery data, 217 hypothyroid myopathy, 261-262 hypotonia collagen-related myopathies, 194 DM1, 184 nemaline myopathy, 241-242 neonatal, 29-146, 238-239 Pompe disease, 221 RYR1 myopathy, 234-235 SMA1, 85, 86 X-linked myotubular myopathy, 238 IBM see inclusion body myositis ICI see immune checkpoint inhibitor idiopathic brachial plexus neuropathy see neuralgic amyotrophy idiopathic inflammatory myopathies (IIMs), 248-250 management, 250-251 types, 248 IENFD (intra-epidermal nerve fibre density), 112 IgM anti-MAG polyneuropathy/IgM MGUS associated neuropathies, associated conditions, 102 clinical features, 101-103, 103 clinical history/symptoms, 99 diagnosis, 100-101 examination, 99-100 follow-up, 101 management, 103 nerve motor conduction studies, 100 prevalence, 101 IIMs see idiopathic inflammatory myopathies imaging, 42; see also specific imaging modalities motor neuron diseases, 42 myopathies, 44-46, 45 neuropathies, 42, 43, 44

POEMS syndrome, 105

examination, 163-164

immune-mediated necrotizing

diagnosis, 164

follow-up, 164 management, 165

immune checkpoint inhibitor (ICI)-

clinical history/symptoms, 163

related myasthenia gravis, 164

myopathy (IMNM), 255-256, 265

277

examination, 206



Cambridge University Press & Assessment 978-1-108-74418-8 — Neuromuscular Disease 2nd Edition Jessica E. Hoogendijk, Marianne de Visser, Pieter A. van Doorn, Erik H. Niks

Index

More Information

immune-mediated necrotizing	comparison with myasthenia gravis,
myopathy (IMNM), (cont.)	167
clinical features, 249	development of symptoms, 167
clinical history/symptoms, 252	diagnosis, 166
diagnosis, 252–253	electrodiagnostic studies, 38, 40
examination, 252	examination, 166
follow-up, 253–254, 254	follow-up, 166
imaging, 45 in juveniles, 246	management, 168 laminopathies, imaging, 45
management, 255	lamins (nuclear envelope proteins), 200
MRI scan, 254	Lasègue sign, Lyme radiculopathy, 141
muscle biopsy, 253	late-onset Pompe disease (LOPD),
statins, 254	219–222, 221
videos, 273	LEMS see Lambert-Eaton myasthenic
immunohistochemical stains, ERN-	syndrome
EURO-NMD recommendations,	leprosy (Mycobacterium leprae), 143
49	clinical history/symptoms, 142, 142
immunotherapy, management of	diagnosis, 142-143
neuromuscular disorders, 57-58	differential diagnoses, 142
inclusion body myositis (IBM), 208, 248,	examination, 142
257–259	follow-up, 143
clinical features, 249, 257	skin biopsy, 143
clinical history/symptoms, 256–257,	limb girdle muscular dystrophies
258-259	(LGMDs), 188–189
diagnosis, 257, 259–260	clinical history/symptoms, 187
examination, 257	CT scan, 188
follow-up, 257	diagnosis, 187 dystrophin–glycoprotein complex
imaging, 45 management, 260	(DGC, 190
muscle atrophy, 258–259	examination, 187
muscle biopsy, 258	follow-up, 188
videos, 273	imaging, 45
infantile-onset Pompe disease (IOPD),	pregnancy and obstetrics, 64
221, 223; see also paediatrics	scapula winging, 187
intensive care unit (ICU)-acquired	subtypes, 188, 189, 203; see also below
weakness, 136-137; see also critical	videos, 272
illness polyneuropathy/myopathy	limb girdle muscular dystrophy R1,
intra-epidermal nerve fibre density	189–190, 190
(IENFD), 112	limb girdle muscular dystrophy R9, 192
Isaac syndrome, 37–38, 123 , 124	clinical features, 192
	clinical history/symptoms, 191
joint contractures	diagnosis, 191
differential diagnoses, 28–29	differential diagnoses, 191
examination, 9	examination, 191
juvenile dermatomyositis (JDM),	follow-up, 191
244-245	limbic encephalitis, 123 LGMDs see limb girdle muscular
clinical history/symptoms, 243, 244 diagnosis, 243–244, 245–246	dystrophies
examination, 243	LOPD (late-onset Pompe disease),
follow-up, 244	219–222, 221
management, 246	LPIN1 deficiency, 33-34
MRI scan, 245	lung weakness see respiratory muscle
muscle biopsy, 244, 245	weakness
juvenile immune-mediated necrotizing	Lyme radiculopathy/Lyme disease, 141
myopathy (IMNM), 246	clinical features in children, 141
	clinical history/symptoms, 139, 140
KCNJ2 gene, 213, 215	diagnosis, 140-141
Kearns-Sayre syndrome (KSS), 232	examination, 139-140
Kennedy disease see spinal and bulbar	follow-up, 141
muscular atrophy	MRI scan, 140
lactate dehydrogenase A deficiency	malignant hyperthermia, 213, 235–237,
(GSDXI, LDHA), 33	268, 270
LAMB2 gene, 170	anaesthetic use, 60-62
Lambert-Eaton myasthenic syndrome	exertional rhabdomyolysis, 270
(LEMS), 3, 166–168	RYR1 myonathy, 236, 237

```
man-in-the-barrel see flail arm
    syndrome
management of neuromuscular
     disorders, 57; see also under specific
     conditions
  anaesthetics, 60-63
  cardiac involvement, 59
  care transitions throughout life
     course, 65-66
  gastrointestinal involvement, 59-60
  genetic counselling, 65
  genetic treatments, 57
  immunotherapy, 57-58
  multidisciplinary care, 58-59
  pain management, 60, 61
  POEMS syndrome, 107
  pregnancy and obstetrics, 63-65, 64
  rehabilitation and palliative care, 58
  respiratory muscle weakness, 59
  surgery, 60-63
  telemedicine, 66
MC (myotonia congenita), 213, 215
McArdle disease, 225
  clinical features, 225
  clinical history/symptoms, 224
  diagnosis, 224-225
  follow-up, 225
  muscle biopsy, 224
  rhabdomyolysis, 225, 225-226
McLeod syndrome, 32
Marcus Gunn ptosis, 16
Marinesco-Sjögren syndrome, 33-34
Medical Research Council scale for
     assessment of muscle strength, 11
MELAS (mitochondrial
     encephalomyopathy with lactic
     acidosis and stroke-like episodes),
     232
MEMSA see myoclonic epilepsy,
     myopathy, sensory ataxia
MERRF see myoclonus epilepsy with
    ragged red fibres
MFMs see myofibrillar myopathies
MFN2 gene, 149
MFS see Miller-Fisher syndrome
MG see myasthenia gravis
MGUS (monoclonal gammopathy of
     undetermined significance)
     polyneuropathy see IgM anti-MAG
     polyneuropathy
microscopic polyangiitis (MPA), 109
Miller-Fisher syndrome (MFS), 96
  MFS-GBS overlap syndrome, 96, 272
mitochondrial DNA (mtDNA) testing,
mitochondrial encephalomyopathy with
     lactic acidosis and stroke-like
     episodes (MELAS), 232
mitochondrial myopathies, 230, 232; see
     also primary mitochondrial
     disorders
```

mitochondrial neurogastrointestinal encephalomyopathy (MNGIE), mitochondrial trifunctional protein (MTP) deficiency/ LCHAD

Miyoshi myopathy, 202-203, 204, 205

deficiency, 33-34

Malignant Hyperthermia (MH) Group,

237

clinical history/symptoms, 165-166,



Cambridge University Press & Assessment 978-1-108-74418-8 — Neuromuscular Disease 2nd Edition

Jessica E. Hoogendijk , Marianne de Visser , Pieter A. van Doorn , Erik H. Niks

Index

More Information

Index

MLPA (multiplex ligation-dependent normal muscle tissue, 48 myotonic dystrophies; see also below rimmed vacuoles, 207-208, 208 probe amplification), 54 imaging, 45 MMN see multifocal motor neuropathy RYR1 myopathy, 235 videos, <u>273</u> myotonic <u>dystrophy</u> type 1 (DM1), stains, 47, 49 modified Erasmus GBS Respiratory Insufficiency Score (mEGRIS), 94, techniques and tissue preparation, 48 183-185 muscle chloride channelopathy, 212 clinical features, 184, 186 Moebius syndrome, 16, 17 muscle contractures clinical history/symptoms, 182, 183 differential diagnoses, 27 monoclonal gammopathy of diagnosis, 182-183 undetermined significance examination, 9 examination, 182 (MGUS) see IgM anti-MAG muscle glycolysis disorders, 224; see also facial appearance, 183 McArdle disease polyneuropathy follow-up, 183 muscle hypertrophy, differential Morvan syndrome, 123, 124 management, 184 diagnoses, 28 clinical history/symptoms, 122 myotonic dystrophy type 2 (DM2), muscle ultrasound (MUS), 42 diagnosis, 122-123 185-186 muscular dystrophies, 190; see also electrodiagnostic studies, 37-38 clinical features, 186, 186 Becker muscular dystrophy; examination, 122 clinical history/symptoms, 185 Duchenne muscular dystrophy; follow-up, 123 diagnosis, 185 Emery-Dreifuss muscular videos, 273 examination, 185 dystrophy; limb girdle muscular motor neuron diseases, 1; see also follow-up, 185 dystrophies phenotypes, 186 amyotrophic lateral sclerosis; MuSK-myasthenia gravis, 162-163 primary lateral sclerosis; myotubular myopathy see X-linked clinical features, 162 progressive muscular atrophy myotubular myopathy clinical history/symptoms, 161 differential diagnoses, 35 diagnosis, 162 electrodiagnostic studies, 35 NA see neuralgic amyotrophy imaging, 42, 45 examination, 161-162 NARP (neuropathy, (episodic) ataxia MR (magnetic resonance) imaging, 42 follow-up, 162 and retinitis pigmentosa), 232 management, 163 NEB gene, 241 EDMD, 199 tongue abnormalities, 162 nemaline myopathy, 241-243 GNE myopathy, 207 myalgia and cramps syndrome, 178 HSAN, 152 clinical features, 242 myasthenia gravis (MG), 2-3; see also hyperCKaemia, 267 clinical history/symptoms, 240 AChR-myasthenia gravis; drug-IMNM, 254 diagnosis, 241 induced myasthenia gravis; differential diagnoses, 241 JDM, 245 Lyme radiculopathy, 140 immune checkpoint inhibitorexamination, 241 related myasthenia gravis; MuSKfollow-up, 241 MMN, 121 myasthenia gravis segmental SMA, 83 rods in muscle fibres, 241, 242 comparison with LEMS, 167 MTM 1 myopathy, see X-linked TPM2 gene, 242 electrodiagnostic studies, 38, 40 myotubular myopathy neonatal hypotonia, differential multidisciplinary care, 58-59 examination, 11-13 diagnoses, 29-30 fatigability tests, 13 multifocal motor neuropathy (MMN), neonatal myasthenia gravis, 16, 17 pregnancy and obstetrics, 64 118, 120-122 neostigmine test, 13 clinical history/symptoms, 118 videos, 273 nerve biopsy, 48-50 diagnosis, 118-119, 120 myasthenic crisis, 161 IgM anti-MAG polyneuropathy, 103 Mycobacterium leprae see leprosy vasculitic neuropathy, 108, 108, 109 diagnostic criteria, 119 myoclonic epilepsy, myopathy, sensory differential diagnoses, 120 nerve motor conduction studies, IgM ataxia (MEMSA), 232 examination, 118 anti-MAG polyneuropathy, 100 follow-up, 120 myoclonus epilepsy with ragged red nerve pain, management, 60 imaging, 42, 44 fibres (MERRF), 232, 233 nerve ultrasound (NUS), 42 immunotherapy, 57 myofibrillar myopathies (MFMs), 208, neuralgic amyotrophy (NA), 19, 210-211 125-126 MRI scan, 121 clinical features, 211 clinical features, 126 multi/minicore myopathy, 16 clinical history/symptoms, 209 multiminicore disease, 19-20 clinical history/symptoms, 124-125, diagnosis, 209 multiple acyl-coenzyme 125 A dehydrogenase deficiency examination, 209 diagnosis, 125 (MADD), 33-34 follow-up, 209-210 examination, 125 muscle biopsy, 210 multiplex ligation-dependent probe hereditary, 126-127 amplification (MLPA), 54 myokymia management, 126 differential diagnoses, 27 muscle(s), action and innervation, 11, 12 prognosis, 126 examination, 8 muscle biopsy, 47, 48 neurofilament light chain, 79 myosin myopathy (MyHC IIa), 16 BMD, 177, 179 neuromuscular junction disorders, 2-3; myositis see idiopathic inflammatory see also myasthenia gravis DM, 249-250 myopathies; immune-mediated Dok7 myasthenic syndrome, 169 electrodiagnostic studies, 38, 40 necrotizing myopathy drug-induced myopathies, 263 therapies, 3 myositis-associated antibodies (MAAs), endocrine myopathy, 261 neuronopathies, 2, 25, 26, 37; see also IBM, 258 sensory neuronopathies myotonia IMNM, 253 neuropathic hereditary transthyretin differential diagnoses, 27 IDM, 244, 245 (TTR) amyloidosis, 155, 156 examination, 8-9 McArdle disease, 224 neuropathic pain, 2 myotonia congenita (MC), 213, 214, 215 MFM, 210 neuropathy, (episodic) ataxia and MTM1 myopathy, 239 myotonic discharges, EMG, 271-272 retinitis pigmentosa (NARP), 232



Cambridge University Press & Assessment 978-1-108-74418-8 — Neuromuscular Disease 2nd Edition Jessica E. Hoogendijk , Marianne de Visser , Pieter A. van Doorn , Erik H. Niks

More Information

Index

neutral lipid storage disease, 30-31, 32 next-generation sequencing (NGS) panels, 53-54; see also genetic testing Nonaka myopathy see GNE myopathy nondystrophic myotonias, 212, 214, 215 nuclear envelope proteins, 200 nutritional deficiencies, myopathies, 263 obstetrics, management, 63-65, 64 oculopharyngeal muscular dystrophy (OPMD), 196-197, 208 clinical features, 197 clinical history/symptoms, 196 diagnosis, 196 examination, 196 management, 197 oculopharyngodistal myopathy, 16, 18 ophthalmoplegia, external, 9, 16 overlap-myositis, clinical features, 249 PABPN1 gene, 196-197 paediatrics; see also juvenile dermatomyositis care transitions throughout life course, 65-66 examination, 7-14 immune-mediated necrotizing myopathy, 246 Lyme radiculopathy, 141 Pompe disease, 221, 223 differential diagnoses, 28 management, 60 treatment algorithm, 61 palliative care, 58 ALS, 76 SMA type 1, 87 PAN see polyarteritis nodosa paramyotonia congenita, 213 paraneoplastic sensory neuronopathy, 115 antibodies associated with cancer in, 116 clinical history/symptoms, 114 diagnostic criteria, 115 examination, 114 follow-up, 114 ganglionopathies, 114 Parsonage-Turner syndrome. see neuralgic amyotrophy percussion-induced muscle mounding (PIMMs), caveolinopathy, 201 percussion-induced rapid contractions (PIRCs), caveolinopathy, 201 peripheral nerve hyperexcitability syndromes, 123, 123. see also Isaac syndrome; limbic encephalitis; Morvan syndrome; rippling muscle disease; stiff person syndrome electrodiagnostic studies, 37-38 spectrum of disorders, 124 videos, 272-273 PET (positron emission tomography), POEMS syndrome, 105 PGT (pre-implantation genetic testing), phosphatidic acid phosphatase deficiency, 33-34

phosphofructokinase deficiency (Tarui disease/GSDVII), 33 phosphoglycerate kinase deficiency (PGK1), 33 phosphoglycerate mutase deficiency (GSDX), 33 Pierson syndrome, 170 Plexopathy, electrodiagnostic studies, 37 PLS see primary lateral sclerosis PMA see progressive muscular atrophy PMDs see primary mitochondrial disorders PMP-22 gene, CMT, 144, 146-147 HNPP, **145–146**, 147–148 POEMS see polyneuropathy, organomegaly, endocrine manifestations, monoclonal protein, and skin changes polio-like syndrome, West Nile virus, 93 poliomyelitis anterior acuta, 1, 91; see also postpolio syndrome polyarteritis nodosa (PAN), 109 polyneuropathies, 2; see also chronic polyneuropathy demyelinating features, 38 electrodiagnostic studies, 37, 38, 39 risk factors, 134 polyneuropathy, organomegaly, endocrine manifestations, monoclonal protein, and skin changes (POEMS), 105-107 clinical history/symptoms, 104, 106 diagnosis, 104-105 diagnostic criteria, 106 examination, 104 follow-up, 105 management, 107 muscle atrophy and skin discolouration, 104, 105 PET imaging, 105 prognosis, 107 polyradiculopathy, electrodiagnostic studies, 37 Pompe disease, 219-222 clinical features, 221 clinical history/symptoms, 218, 218, 221 diagnosis, 218-219 examination, 218 follow-up, 219 imaging, 45 management, 223 muscle weakness distribution, 220 pregnancy and obstetrics, 64 respiratory muscle weakness, 222 videos, 273 postpolio syndrome (PPS), 92-93, 92 clinical history/symptoms, 91 diagnosis, 1-2, 91 diagnostic criteria, 93 examination, 91 follow-up, 91 postural tremor, differential diagnoses, 27 PP (primary periodic paralyses), 213, 215 PPS see postpolio syndrome prednisolone/prednisone, 157, 160, 162

pregnancy, 63-65, 64 pre-implantation genetic testing (PGT), 65 primary lateral sclerosis (PLS), 40-41, 79 anterior horn cell diseases, 1 clinical history/symptoms, 1, 77, 80 diagnosis, 77 diagnostic criteria, 77, 79 differential diagnoses, 78 examination, 77 follow-up, 77 frequency of disease, 79 prognosis, 80 primary mitochondrial disorders (PMDs), 230-231 clinical features, 231, 233 with neuromuscular features, 232 primary periodic paralyses (PP), 213, 215 progressive muscular atrophy (PMA), 1, 80 - 81clinical history/symptoms, 1, 79 diagnosis, 1-2, 80 examination, 79-80 flail arm syndrome, 81 follow-up, 80 frequency of disease, 80 pseudobulbar palsy, 272 ptosis, examination, $\overline{9}$ PURA gene, 170 pyroxidine deficiency, 138 quadriceps sparing myopathy see GNE myopathy Rab7 gene, 150, 150-151 ragged red fibres, mitochondrial myopathies, 233; see also myoclonus epilepsy with ragged red fibres RAPSN gene, 170 repetitive nerve stimulation (RNS), 171 respiratory muscle weakness differential diagnoses, 19 examination, 10 JDM, 246 management, 59 Pompe disease, 222 reversible infantile respiratory chain deficiency (RIRCD), 232 rhabdomyolysis (RML), 270; see also exertional rhabdomyolysis CPT2 deficiency, 227-228 differential diagnoses, 32-34 drug-induced myopathies, 263, 265 McArdle disease, 225, 225-226 mitochondrial disorders, 232 RYR1 myopathy, 237 riboflavin transporter deficiency (RTD), 16, 18 riluzole, 76 rimmed vacuoles, muscle biopsy,

207-208, 208; see also GNE

myopathy

rippling muscles, 201

examination, 9

caveolinopathy, 200-201

differential diagnoses, 27



Cambridge University Press & Assessment 978-1-108-74418-8 — Neuromuscular Disease 2nd Edition

Jessica E. Hoogendijk , Marianne de Visser , Pieter A. van Doorn , Erik H. Niks

Index

More Information

Index

SFN. 111 rippling muscle disease, 123; see also stiff person syndrome, 123 skin discolouration, POEMS syndrome, caveolinopathy stiffness, differential diagnoses, 28 RIRCD (reversible infantile respiratory 104, 105 STIR see short tau inversion recovery skin lesions, vasculitic neuropathy, 108 chain deficiency), 232 surgical options, 60-63 skin rash, JDM, 243, 244 RML see rhabdomyolysis swallowing difficulties, management, SMA see spinal muscular atrophy RNS (repetitive nerve stimulation), 171 59-60 small-cell lung cancer (SCLC), 166-168, rods, in muscle fibres, 241, 242; see also nemaline myopathy TANGO2-syndrome, 33-34 small-fibre neuropathy (SFN), 111-113 Tarui disease, 33 RYR1 gene, 269 RYR1 myopathy, 235-237 associated conditions, 111 telemedicine, 66 causality, 113 clinical features, 236 tendon reflexes, 13-14 clinical history/symptoms, 110 clinical history/symptoms, 234, 234 therapy see management of diagnosis, 110-111, 112-113 diagnosis, 234-235 neuromuscular disorders differential diagnoses, 111-112 examination, 234 thiamine deficiency, alcoholic examination, 110 follow-up, 235 polyneuropathy, 131 follow-up, 111 thymidine kinase 2 deficiency, 33-34 muscle biopsy, 235 grading, 112 thyroid-associated ophthalmopathy, 262 types, 236, 236 intra-epidermal nerve fibre density, thyrotoxic hypokalaemic periodic 112 paralysis (TPP), 213 SAAM (statin-associated autoimmune management, 113 tick bites, vasculitic neuropathy, 108; see myopathy), 265 phenotypes, 113 SANDO (sensory ataxic neuropathy, also Lyme disease/Lyme skin biopsy, 111 dysarthria, ophthalmoplegia), 232 radiculopathy symptoms suggesting, 110 sarcoid myopathy, 21-22, 249-250 time course of disease, history taking, 5 sarcoglycanopathies, 30-31, 32-34, 59, small interfering ribonucleic acids tongue abnormalities (siRNAs), 57 differential diagnoses, 28 227 small polyphasic motor unit potentials, SBMA see spinal and bulbar muscular examination, 10 EMG, 271-272 MuSK-myasthenia gravis, 162 atrophy SMPX distal myopathy, 208 TPM2 gene, 242 scapular winging sodium channel myotonia, 213 differential diagnoses, 20 TPP (thyrotoxic hypokalaemic periodic examination, 10 spastic paraplegia, differential diagnoses, paralysis), 213 LGMD, 187 transthyretin amyloidosis (TTR), 155, spasticity of gait, 272 Lyme radiculopathy, 140 NA, 125 spasticity, management, 60 clinical history/symptoms, 154 spinal and bulbar muscular atrophy diagnosis, 154 SCLC see small-cell lung cancer (SBMA), 1, 84-85, 85 SCN4A gene, 213 differential diagnoses, 154 clinical history/symptoms, 84, 85 segmental muscular atrophy, 1, 82, examination, 154 diagnosis, 1-2, 84 follow-up, 154-155 clinical history/symptoms, 82 examination, 84 genetic treatments, 57 follow-up, 84 neuropathic type, 155, 156 diagnosis, 1-2, 82 differential diagnoses, 82 videos, 272 transthyretin familial amyloid spinal muscular atrophy (SMA), 1; see polyneuropathy see hereditary examination, 82 also segmental muscular atrophy follow-up, 83 transthyretin amyloidosisand see below MRI scan, 83 polyneuropathy diagnosis, 1-2 therapies, 2 transthyretin (TTR) transport protein, genetic treatments, 57 sensory ataxic neuropathy, dysarthria, 155 imaging, 45 ophthalmoplegia (SANDO), 232 treatments see management of pregnancy and obstetrics, 64 sensory neuronopathies (SNN), neuromuscular disorders 114-115; see also paraneoplastic therapies, 2 tremor, differential diagnoses, 27 spinal muscular atrophy (SMA) type 1, Trendelenburg sign sensory neuronopathy Lyme radiculopathy, 140 antibodies associated with cancer in clinical history/symptoms, 85 SNN, 116 Pompe disease, 218 diagnosis, 86 diagnostic criteria, 115 TTR amyloidosis see transthyretin differential diagnoses, 86 serum creatine kinase see creatine kinase amyloidosis SFN see small-fibre neuropathy examination, 85-86 TTR (transthyretin) transport protein, follow-up, 86 shawl sign, 247 single-fibre electromyography (SfEMG), management, 87 spinal muscular atrophy (SMA) type 3, 38 Ullrich congenital muscular dystrophy 89-90 skeletal muscle channelopathies, 213, (UCMD), 45, 194, 194, 195 classificatory features, 89 214-215 ultrasound (US), 42; see also muscle clinical history/symptoms, 88 ultrasound; nerve ultrasound clinical history/symptoms, 212 diagnosis, 88-89 diagnosis, 212 Dok7 myasthenic syndrome, 170 examination, 88 videos, <u>272</u> examination, 212 follow-up, 89 follow-up, 214 types, 213 management, 90 vaccine-derived poliovirus (VDPV), 93 phenotypes, 90 Valsalva manoeuvre, 14 skin abnormalities statin-associated autoimmune DM, 250 vascular endothelial growth factor myopathy (SAAM), 265 examination, 8 (VEGF), 105 statins, 254, 263, 264, 264-265 skin biopsy, 50-51 vasculitic neuropathy, 109-110 leprosy, 143 steroids see corticosteroids classificatory features, 109



Cambridge University Press & Assessment 978-1-108-74418-8 — Neuromuscular Disease 2nd Edition Jessica E. Hoogendijk , Marianne de Visser , Pieter A. van Doorn , Erik H. Niks Index <u>More Information</u>

Index

vasculitic neuropathy (cont.) clinical history/symptoms, 107, 108, 109 diagnosis, 108, 109 examination, 107-108 follow-up, 108-109 management, 109-110 nerve biopsy, 108, 108 VCP distal myopathy, 208 ventilator weaning, ICU-acquired weakness, 136-137 very long-chain acyl-CoA dehydrogenase deficiency (VLCAD), 33-34, 229 video fluoroscopic swallow studies (VFSS), 10 videos case studies, 272-273 EMG, 271-272 ultrasound, 272

vitamin B1 deficiency, alcoholic polyneuropathy, 131 vitamin B6 deficiency, 138 vitamin B12 deficiency, **78** vitamin D deficiency, 263, **264**, 266 voltage-gated calcium channels, 3

Wartenberg migrant sensory neuropathy, 117–118 clinical features, 117 clinical history/symptoms, 116–117 diagnosis, 117 examination, 117 follow-up, 117 Welander distal myopathy, 208 West Nile virus (WNV), 93; see also postpolio syndrome Western blot analysis, 177 whole exome-sequencing (WES), winging, scapula see scapula winging

X-linked dilated cardiomyopathy (DCM), 178, 179 X-linked muscular dystrophy see Emery-Dreifuss muscular dystrophy X-linked myotubular myopathy (MTM1), 239-240 arachnodactyly, 238 clinical history/symptoms, 238, 239 diagnosis, 238-239 examination, 238 follow-up, 239 muscle biopsy, 239 X-linked recessive diseases. see Becker muscular dystrophy; Duchenne

muscular dystrophy