Table S2. Clinical features and the impact of genetic diagnosis on clinical management.

Patient ID	Sex	AAO	Clinical features	Conventional investigation	Disease gene (phenotype /MIM number)	MOI	Impact on clinical management				
							Investigation	Treatment	Surveillance	Family counseling	Prognostication
Group 1: Inherited myopathy											
CM1	M	8Y6M	• Back pain and difficulty squatting • Proximal muscle weakness • Rigid spine, scoliosis • Scapular winging	• CK 1931 U/L • MLPA for DMD: negative • Edx: irritable myopathy • Muscle biopsy: myofibrillar myopathy	***FHL1*** (Myopathy, X-linked, with postural muscle atrophy/ 300696)	XR	-	-	Cardiac and respiratory involvement	• Family testing (father, mother)	Variable
CM2	F	27D	• Dilated cardiomyopathy • Delay motor milestone • Proximal muscle weakness	• Plasma amino acid: NL • Urine organic acid: NL	***MYH7*** (Cardiomyopathy, dilated, 1S/ 613426)	AD	Avoid muscle biopsy	-	Hypertrophic and dilated cardiomyopathy	• Family testing (father, mother)	Yes
CM3*	F	4Y	• Distal weakness • Scoliosis • Mild restrictive lung disease	• CK 591 U/L • Edx: myopathic change	***MYH7*** (Laing distal myopathy/ 160500)	AD	-	-	Hypertrophic and dilated cardiomyopathy	• Family testing (mother)	Yes
CM4*	M	4Y6M	• Distal weakness • Scoliosis • Moderate restrictive lung disease	• CK 608 U/L • Muscle biopsy: myopathic change • Edx: myopathic change	***MYH7*** (Laing distal myopathy/ 160500)	AD	-	-	Hypertrophic and dilated cardiomyopathy	• Family testing (mother)	Yes
CM5	M	1Y	• Delay motor milestone • Proximal muscle weakness • Myopathic face • Scoliosis • Chronic respiratory insufficiency	• CK 70 U/L • Muscle biopsy: nemaline myopathy	***NEB*** (Nemaline myopathy 2/ 256030)	AR	-	-	Respiratory and bulbar involvement	• Family testing (father, mother)	Yes
CM6	M	3M	• Severe scoliosis • Malignant hyperthermia • Myopathic face • Proximal muscle weakness	• CK 279 U/L • Muscle biopsy: tubular aggregate myopathy • Edx: myopathic change	***RYR1*** (Congenital myopathy 1B/ 255320)	AR	-	-	Respiratory involvement	• Family testing (father, mother) • Malignant hyperthermia risk	Yes
CM7	M	3M	• Delay head up • Ptosis with opthalmoplegia • Proximal musle weakness	• CK 148 U/L • Anti-AchR: positive • Edx: NL, RNS: NL	***RYR1*** (Congenital myopathy 1B/ 255320)	AR	Avoid muscle biopsy	Off immunosuppressive drugs (previously treated as MG)	Respiratory involvement	• Family testing (father, mother) • Malignant hyperthermia risk	Yes
CM8	M	birth	• Hypotonia • Elongated face, high arch palate • Ptosis with opthalmoplegia • Undescended testes • Oromotor dysfunction	• CK 58 U/L • Anti-AchR: negative • Muscle biopsy: myopathic change with type 1 fiber predominance • Edx: myopathic, RNS: negative	***RYR1*** (Congenital myopathy 1B/ 255320)	AR	-	-	Respiratory involvement	• Family testing (father, mother) • Malignant hyperthermia risk	Yes
CM9	M	birth	• Hypotonia • Myopathic face • Ptosis with opthalmoplegia • Proximal musle weakness	• CK 116 U/L, lactate 1.5 mmol/L	***RYR1*** (Congenital myopathy 1B/ 255320)	AR	Avoid muscle biopsy	-	Respiratory involvement	• Malignant hyperthermia risk	Yes
CM10	M	8Y9M	• Walking difficulty • Proximal muscle weakness	• CK 95 U/L • Muscle biopsy: non specific myopathic change	***RYR1*** (Congenital myopathy 1B/ 255320)	AD/AR	-	-	Respiratory involvement	• Family testing (mother) • Malignant hyperthermia risk	Yes
CM11	F	12Y9M	• Severe scoliosis • Facial weakness, high arch palate • Proximal muscle weakness • Rigid spine • Moderate restrictive lung disease	• CK 168 U/L • Edx: myopathic change	***SELENON*** (Congenital myopathy 3 with rigid spine/ 602771)	AR	Avoid muscle biopsy	-	Cardiac and respiratory involvement	-	Yes

CM12	M	birth	• Hypotonia, respiratory failure • Finger flexion contracture • Myopathic face • Proximal muscle weakness • Rigid spine • Scoliosis • Moderate restrictive lung disease • Abernethy malformation	• CK 86 U/L • Muscle biopsy: non-specific change	***TTN*** (Limb-girdle muscular dystrophy/ 608807)	AR	-	-	Cardiac and respiratory involvement	• Family testing (father, mother)	Yes
CM13	M	birth	• Hypotonia, respiratory failure • Myopathic face • Proximal muscle weakness • Oromotor dysfunction	• CK 57 U/L • Muscle biopsy: myopathic change	***TTN*** (Limb-girdle muscular dystrophy/ 608807)	AR	-	-	Cardiac and respiratory involvement	• Family testing (father, mother)	Yes
CM14	F	birth	• Arthrogryposis multiplex congenita • Proximal muscle weakness • Oromotor dysfunction • Severe scoliosis • Dilated cardiomyopathy	• CK 37 U/L	***TTN*** (Congenital myopathy 5 with cardiomyopathy/ 611705)	AR	Avoid muscle biopsy	-	Cardiac and respiratory involvement	• Family testing (father, mother)	Yes
CM15	M	6M	• Delay motor milestone • Facial weakness • Hypotonia	• CK 243 U/L • MLPA for *SMN1*: negative	19q13.33q13.41 deletion	-	-	-	-	-	-
DMD1	M	3Y6M	• Proximal muscle weakness (LE) • Calf pseudohypertrophy • Restrictive lung disease	• CK 18930 U/L • MLPA for *DMD*: negative	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	Avoid muscle biopsy	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
DMD2	M	3Y	• Frequent falling • Proximal muscle weakness (LE) • Calf pseudohypertrophy	• CK 12704 U/L • MLPA for DMD: negative	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	Avoid muscle biopsy	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	• Family testing (mother) • Cardiac surveillance in DMD-carriers	Yes
DMD3	M	7Y	• Frequent falling • Proximal muscle weakness (LE) • Calf pseudohypertrophy	• CK 15097 U/L • MLPA for DMD: negative • Muscle biopsy: dystrophinopathy	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	-	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
DMD4	M	NA	• Down syndrome • Unexplained transaminitis • Severe intellectual disability • Proximal muscle weakness (LE) • Calf pseudohypertrophy	• CK >22000 U/L, AST, ALT • MLPA for DMD: negative • Muscle biopsy: dystrophinopathy	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	-	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	• Family testing (mother) • Cardiac surveillance in DMD-carriers	Yes
DMD5	M	4Y	• Tiptoe walking • Proximal muscle weakness (LE) • Calf pseudohypertrophy	• CK 15645 U/L • MLPA for DMD: negative	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	Avoid muscle biopsy	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
DMD6	M	3Y6M	• Walking difficulty • Proximal muscle weakness (LE) • Ankle contractures	• CK 9196 U/L • MLPA for DMD: negative	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	Avoid muscle biopsy	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
DMD7	M	5Y	• Frequent falling • Proximal muscle weakness (LE) • Calf pseudohypertrophy	• CK 16571 U/L • MLPA for DMD: negative	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	Avoid muscle biopsy	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
DMD8	M	1Y3M	• Hypomelanosis of Ito • Macrocephaly • Delayed walking • Proximal muscle weakness (LE)	• CK 16836 U/L • MLPA for DMD: negative • Muscle biopsy: dystrophinopathy • Brain MRI: NL	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	-	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	• Family testing (mother) • Cardiac surveillance in DMD-carriers	Yes
DMD9	M	5Y	• Proximal muscle weakness (LE) • Calf pseudohypertrophy • Severe obstructive sleep apnea	• CK 12139 U/L • MLPA for DMD: negative	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	Avoid muscle biopsy	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	• Family testing (mother) • Cardiac surveillance in DMD-carriers	Yes

DMD10	M	4Y	• Frequent falling • Proximal muscle weakness (LE) • Calf pseudohypertrophy	• CK 7708 U/L • MLPA for DMD: negative	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	Avoid muscle biopsy	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
DMD11	M	5Y	• Frequent falling • Proximal muscle weakness (LE) • Calf pseudohypertrophy • Scoliosis	• CK 9874 U/L • MLPA for DMD: negative • Muscle biopsy: dystrophinopathy	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	-	• Steroid • ACEI/ARB • Candidate for Ataluren	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
DMD12	M	3Y	• Waddling gait • Proximal muscle weakness (LE) • Calf pseudohypertrophy	• CK 9089 U/L • MLPA for DMD: negative	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	Avoid muscle biopsy	• Steroid • ACEI/ARB • Candidate for Ataluren	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	• Family testing (mother) • Cardiac surveillance in DMD-carriers	Yes
DMD13	M	4Y	• Tiptoe walking • Proximal muscle weakness (LE) • Calf pseudohypertrophy • Delay speech	• CK 7129 U/L • MLPA for DMD: negative • Muscle biopsy: dystrophinopathy	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	-	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
DMD14	M	2Y	• Tiptoe walking • Proximal muscle weakness (LE) • Calf pseudohypertrophy	• CK 13661 U/L • MLPA for DMD: negative • Muscle biopsy: dystrophinopathy	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	-	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
DMD15	M	4Y	• Tiptoe walking • Proximal muscle weakness (LE) • Calf pseudohypertrophy • Ankle contractures	• CK 5848 U/L • MLPA for DMD: negative • Muscle biopsy: end stage muscle disease	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	-	• Steroid • ACEI/ARB	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	• Family testing (mother) • Cardiac surveillance in DMD-carriers	Yes
DMD16	M	1Y3M	• Frequent falling • Proximal muscle weakness (LE) • Calf pseudohypertrophy	• CK 13287 U/L • MLPA for DMD: negative	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	Avoid muscle biopsy	• Steroid • ACEI/ARB • Candidate for Ataluren	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
DMD17	M	1Y3M	• Frequent falling • Proximal muscle weakness (LE) • Calf pseudohypertrophy • GDD, ADHD, autism • Epilepsy	• CK 10247 U/L • MLPA for DMD: negative	***DMD*** (Duchenne muscular dystrophy/ 310200)	XR	Avoid muscle biopsy	• Steroid • ACEI/ARB • Candidate for Ataluren	• Dilated cardiomyoathy • Respiratory involvement • Scoliosis	-	Yes
MD1	M	1Y3M	• Delay walking • Proximal muscle weakness • GDD • Scoliosis	• CK 1279 U/L • MLPA for DMD: negative	***CHKB*** (Muscular dystrophy, congenital, megaconial type/ 602541)	AR	Avoid muscle biopsy	-	Dilated cardiomyopathy	-	Yes
MD2	F	1Y3M	• Delay walking • Proximal muscle weakness • Ichthyosis • Dilated cardiomyopathy since 9Y	• CK 1545 U/L • MLPA for DMD: negative	***CHKB*** (Muscular dystrophy, congenital, megaconial type/ 602541)	AR	Avoid muscle biopsy	-	Dilated cardiomyopathy	-	Yes
MD3	F	1Y	• Proximal muscle weakness • Proximal joint contracture • Distal joint hyperlaxity • Keratosis pilaris, keloids	• CK 623 U/L • PCR-RFLP for SMN1: negative • Muscle biopsy: myopathic change, SSCD	***COL6A1*** (Ullrich congenital muscular dystrophy 1A/ 254090)	AD	-	-	Respiratory and cardiac involvement	• Family testing (father, mother)	Yes
MD4	M	4M	• Severe scoliosis • Hip dislocation • Hypotonia • Proximal muscle weakness • Distal joint laxity	• CK 300 U/L • Muscle biopsy: myopathic change, SSCD	***COL6A1*** (Ullrich congenital muscular dystrophy 1A/ 254090)	AD	-	-	Respiratory and cardiac involvement	• Family testing (father, mother)	Yes

MD5	F	1Y	• Hip dislocation • Proximal muscle weakness • Hypertrophic scar • Distal joint laxity	• CK 424 U/L • PCR-RFLP for SMN1: negative • Muscle biopsy: myopathic change, SSCD	***COL6A1*** (Ullrich congenital muscular dystrophy 1A/ 254090)	AD	-	-	Respiratory and cardiac involvement	-	Yes
MD6	M	4Y	• Walking difficulty • Proximal muscle weakness • Proximal joint contracture • Bethlem sign • Distal joint laxity • Keratosis pilaris, keloid • Epilepsy since 16Y	• CK 815 U/L • Edx: myopathic pattern • Muscle biopsy: myopathic change	***COL6A1*** (Bethlem myopathy 1A /158810)	AD	-	-	Respiratory and cardiac involvement	-	Yes
MD7	M	birth	• Hypotonia • Proximal muscle weakness • Proximal joint contracture • Rigid spine • Distal joint laxity • Keratosis pilaris • Scoliosis	• CK 170 U/L • PCR-RFLP for SMN1: negative • Muscle biopsy: myopathic change, complete negative COL6, absent staining on COL6/COL6 double stain	***COL6A2*** (Ullrich congenital muscular dystrophy 1B/ 620727)	AR	-	-	Respiratory and cardiac involvement	• Family testing (father, mother)	Yes
MD8	F	1Y6M	• Left torticollis • Delay motor milestone • Proximal muscle weakness • Distal joint laxity • Keratosis pilaris • Scoliosis	• Neck U/S: very small size of left sternocleidomastoid with predominately tendon component • CK 283 U/L	***COL6A2*** (Ullrich congenital muscular dystrophy 1B/ 620727)	AD	Avoid muscle biopsy	-	Respiratory and cardiac involvement	-	Yes
MD9	F	1Y	• Right sternocleidomastoid (3W) and left torticollis (1Y) • Proximal muscle weakness • Distal joint laxity • Keratosis pilaris • Scoliosis	• CK 563 U/L • Edx: myopathic pattern • Muscle biopsy: myopathic change, faint COL6 staining	***COL6A2*** (Ullrich congenital muscular dystrophy 1B/ 620727)	AD	-	-	Respiratory and cardiac involvement	-	Yes
MD10	F	birth	• Hip dislocation • Proximal muscle weakness • Proximal joint contracture • Hypertrophic scar • Keratosis pilaris • Distal joint laxity	• CK 86 U/L • Muscle biopsy: myopathic change, SSCD	***COL6A3*** (Ullrich congenital muscular dystrophy 1C/ 620728)	AD	-	-	Respiratory and cardiac involvement	-	Yes
MD11	F	4M	• Delay motor milestone • Elongated face • Facial weakness • High arch palate • Proximal muscle weakness	• CK 737 U/L • MLPA for SMN1: negative • Muscle biopsy: myopathic change, focally positive (faint, patchy, partial-deficiency) of merosin stainning • Brain MRI: diffuse and bilateral symmetrical T2 hyperintense white matter lesion with spared corpus collosum and corticospinal tract	***LAMA2*** (Muscular dystrophy, congenital, merosin deficient/ 607855)	AR	Brain MRI	-	Respiratory and cardiac involvement	• Family testing (father, mother)	Yes
MD12	F	birth	• Hypotonia • GDD • Proximal muscle weakness • Pectus excavatum	• CK 3326 U/L • Edx: myopathic pattern • Muscle biopsy: dystrophic change, faint merosin staining • Brain MRI: diffuse and bilateral symmetrical T2 hyperintense white matter lesion with spared corpus collosum and corticospinal tract	***LAMA2*** (Muscular dystrophy, congenital, merosin deficient/ 607855)	AR	Brain MRI	-	Respiratory and cardiac involvement	• Family testing (father, mother)	Yes

MD13*	M	6M	• Delay motor milestone • Hypotonia • Myopathic face • High arch palate • Proximal muscle weakness • Scoliosis • Severe restrictive lung disease	• CK 443 U/L • Edx: myopathic pattern • Muscle biopsy: dystrophic change, absent merosin staining • Brain MRI: diffuse and bilateral symmetrical T2 hyperintense white matter lesion with spared corpus collosum and corticospinal tract	***LAMA2*** (Muscular dystrophy, congenital, merosin deficient/ 607855)	AR	Brain MRI	-	Respiratory and cardiac involvement	-	Yes
MD14*	F	6M	• Delay motor milestone • Hypotonia • Myopathic face • High arch palate • Proximal muscle weakness • Scoliosis • Hip dislocation • Severe restrictive lung disease	• CK 2593 U/L • Edx: myopathic pattern	***LAMA2*** (Muscular dystrophy, congenital, merosin deficient/ 607855)	AR	Avoid muscle biopsy	-	Respiratory and cardiac involvement	-	Yes
MD15	M	3M	• Delayed motor milestone • Hypotonia • Proximal muscle weakness	• CK 1046 U/L • MLPA for SMN1: negative	***LAMA2*** (Muscular dystrophy, congenital, merosin deficient/ 607855)	AR	Avoid muscle biopsy	-	Respiratory and cardiac involvement	-	Yes
MD16	M	5M	• Delayed motor milestone • Hypotonia • Proximal muscle weakness • Focal epilepsy	• CK 1432 U/L • Muscle biopsy: dystrophic change, absent merosin staining • Brain MRI: diffuse and bilateral symmetrical T2 hyperintense white matter lesion with spared corpus collosum and corticospinal tract	***LAMA2*** (Muscular dystrophy, congenital, merosin deficient/ 607855)	AR	Brain MRI	-	Respiratory and cardiac involvement	-	Yes
MD17	M	4M	• Delayed motor milestone • Hypotonia • Proximal muscle weakness • Scoliosis	• CK 6066 U/L • MLPA for SMN1: negative	***LAMA2*** (Muscular dystrophy, congenital, merosin deficient/ 607855)	AR	Avoid muscle biopsy	-	Respiratory and cardiac involvement	• Family testing (father, mother) • Preimplantation genetic testing	Yes
MD18*	F	3Y	• Frequent falling • Proximal muscle weakness • Early contracture • Dilated cardiomyopathy • Atrial fibrillation • Sinus node dysfunction	• Lactate 1.3 mmol/L • Edx: myopathic pattern • Muscle biopsy: dystrophic change	***LMNA*** (Emery-Dreifuss muscular dystrophy 3/ 616516)	AR	-	-	Cardiac conduction defect, dilated or hypertrophic cardiomyopathy	• Family testing (mother) • Cardiac surveillance in carriers	Yes
MD19*	F	3Y	• Frequent falling • Proximal muscle weakness • Early contracture • Dilated cardiomyopathy • Atrial fibrillation	• CK 629 U/L	***LMNA*** (Emery-Dreifuss muscular dystrophy 3/ 616516)	AR	Avoid muscle biopsy	-	Cardiac conduction defect, dilated or hypertrophic cardiomyopathy	• Family testing (mother) • Cardiac surveillance in carriers	Yes
MD20	F	4Y	• Tiptoe walking • Proximal muscle weakness • Early contracture • Rigid spine • Scoliosis	• CK 194 U/L • MLPA for SMN1: negative	***LMNA*** (Emery-Dreifuss muscular dystrophy 2/ 181350)	AD	Avoid muscle biopsy	-	Cardiac conduction defect, dilated or hypertrophic cardiomyopathy	• Family testing (father, mother)	Yes
MD21	F	7M	• Delayed motor milestone • Proximal muscle weakness • Early contracture • Scoliosis • Severe obstructive sleep apnea • Atrial fibrillation	• CK 756 U/L • PCR-RFLP for SMN1: negative • Muscle biopsy: dystrophic change	***LMNA*** (Emery-Dreifuss muscular dystrophy 2/ 181350)	AD	-	-	Cardiac conduction defect, dilated or hypertrophic cardiomyopathy	• Family testing (father, mother)	Yes

MD22	F	2Y	• Tiptoe walking • Proximal muscle weakness • Lumbar lordosis	• CK 3266 U/L • PCR-RFLP for SMN1: negative • Muscle biopsy: dystrophic change	***LMNA*** (Emery-Dreifuss muscular dystrophy 2/ 181350)	AD	-	-	Cardiac conduction defect, dilated or hypertrophic cardiomyopathy	• Family testing (father, mother)	Yes
MM1	F	6Y4M	• Abnormal gait • Proximal weakness • Facial weakness • Hypertrophic cardiomyopathy • Hypoventilation	• CPK 1540 U/L • Alpha-1,4 glucosidase: 0.3 umol/L/h • Edx: irritable myopathy • Muscle biopsy: glycogen accumulation	***GAA*** (Glycogen storage disease 2/ 232300)	AR	-	Candidate for enzyme replacement therapy (Myozyme)	• Cardiomyopathy • Arrhythmia • Diaphragmatic weakness	• Family testing (father, mother, brother)	Yes
MM2	F	birth	• Prematurity, GA 32^{+6} weeks • Polyhydramnios • Profound peripheral hypotonia • Bilateral ptosis • Diaphragmatic paralysis	• CPK 231 U/L • Lactate 1.4 mmol/L, NH_3 32.2 umol/L • *DMPK*: absence of CTG repeat expansion • Muscle biopsy: polyglucosan bodies with a Maltese Cross birefingent pattern under polarized light	***GBE1*** (Glycogen storage disease 4/ 232500)	AR	-	Discontinuation of treatment, extubation	• Cardiomyopathy • Arrhythmia • Hepatic dysfunction • Urinary tract dysfunction	• Family testing (father, mother) • Preimplantation genetic testing	Yes
MC1	M	3Y	• Episodic muscle stiffness • Myotonia with warm-up phenomenon • Muscle hypertrophy	• Edx: myotonic discharge with transient CMAP increase on short exercise test	***CLCN1*** (myotonia congenita, recessive/ 255700)	AR	-	• Lamotrigine • Carbamazepine • Phenytoin • Acetazolamide	-	• Avoid depolarizing muscle relaxants, adrenaline, beta-adrenergic agonists, and propranolol	-
MC2	M	11Y	• Episodic muscle stiffness • Myotonia with warm-up phenomenon • Muscle hypertrophy	-	***CLCN1*** (myotonia congenita, dominant/ 160800)	AD	-	• Lamotrigine • Carbamazepine • Phenytoin • Acetazolamide	-	• Avoid depolarizing muscle relaxants, adrenaline, beta-adrenergic agonists, and propranolol	-
Group 2: Inherited neuropathy											
CMT1	F	birth	• Feet and legs deformities • Distal muscle atrophy • Bilaterak hip and knee flexion contractures • Areflexia	• CPK 113 U/L • Edx:neurogenic change in lower extremities • Brain and spine MRI: NL	***DYNC1H1*** (CMT2O/ 614228)	AD	-	-	-	-	Yes
CMT2	F	2Y	• Delay walking • Distal weakness (LE>UE) • Distal paresthesia (LE) • Scoliosis • Wheelchair-dependent since 7Y	• Edx: diffuse demyelinating sensorimotor polyneuropathy • *PMP22* dup: negative	***EGR2*** (CMT1D/ 607678)	AD	-	-	-	-	Variable
CMT3	M	1Y6M	• Delay walking • Distal weakness (LE>UE) • Distal paresthesia (LE) • Pes cavus, hammer toe, claw hand	• CK 167 U/L • Edx: diffuse demyelinating sensorimotor polyneuropathy • *PMP22* dup: negative	***EGR2*** (CMT1D/ 607678)	AD	-	-	-	-	Variable
CMT4	M	6Y	• Abnormal gait • Distal weakness (LE), hip extensors predominant • No abnormal hair	• Edx: diffuse axonal sensorimotor polyneuropathy • *PMP22* dup: negative	***GAN*** (Giant axonal neuropathy 1/ 256850)	AR	• Brain MRI • Nerve biopsy	Candidate for precision medicine (scAAV9/JeT-GAN)	• Optic atrophy • Ataxia, spasticity, CN dysfunction	• Family testing (father, mother)	-
CMT5	F	8Y10M	• Abnormal gait • Distal weakness (LE) • Rhomberg sign • Pes cavus	• Edx: diffuse intermediate sensorimotor polyneuropathy • CSF: albuminocytologic dissociation	***GDAP1*** (CMT2K/ 607831)	AD	-	Off steroid (previously treated as CIDP)	-	• Family testing (mother)	Yes

CMT6	F	9Y	• Foot deformity (equinovarus) • Distal weakness (LE>UE) • Distal paresthesia (LE)	• Edx: diffuse axonal sensorimotor polyneuropathy • *PMP22* dup: negative	***GDAP1*** (CMT2K/ 607831)	AD	-	-	-	-	Yes
CMT7	F	8Y	• Frequent falling • Distal weakness (LE>UE) • Distal muscle atrophy (LE>UE) • Pes cavus	• CK 99 U/L • Edx: diffuse axonal sensorimotor polyneuropathy • *PMP22* dup: negative	***GDAP1*** (CMT2K/ 607831)	AR	-	-	Vocal cord paresis	• Family testing (mother)	Yes
CMT8	F	6Y	• Frequent falling • Distal weakness (LE>UE) • Distal muscle atrophy (LE>UE) • Pes cavus, hammer toe, foot valgus	• Edx: diffuse axonal sensorimotor polyneuropathy • *PMP22* dup: negative	***GDAP1*** (CMT2K/ 607831)	AD/AR	-	-	-	• Family testing (father, sister)	Yes
CMT9	F	6Y	• Tiptoe walking • Distal weakness (LE>UE) • Distal paresthesia (LE) • Equinovarus	• Edx: diffuse axonal sensorimotor polyneuropathy • *PMP22* dup: negative	***GDAP1*** (CMT2K/ 607831)	AD/AR	-	-	-	• Family testing (father, sister)	Yes
CMT10	M	12Y	• Distal weakness (LE) • Distal paresthesia (LE) • Equinovarus	• Edx: diffuse intermediate sensorimotor polyneuropathy	***GJB1*** (CMTX1/ 302800)	XD	-	-	-	-	Yes
CMT11	F	8Y	• Distal weakness (LE) • Distal paresthesia (LE) • Equinovarus	• Edx: diffuse demyelinating sensorimotor polyneuropathy • *PMP22* dup: negative	***HK1*** (CMT4G/ 605285)	AR	-	-	-	-	-
CMT12	M	1Y	• Never achieved walking • Distal weakness (LE) • Distal paresthesia (LE) • Bilateral hip subluxation	• CK 150 U/L • Brain and spine MRI: NL • Muscle biopsy: neurogenic change • PCR-RFLP for *SMN1*: negative • Edx: diffuse axonal sensorimotor polyneuropathy with secondary demyelination	***IGHMBP2*** (CMT2S/ 616155)	AR	-	-	• Autonomic involvement	-	Yes
CMT13	F	5Y	• Chronic wound on the lateral foot • Distal weakness (LE>UE) • Distal paresthesia (LE>UE) • Equinovarus, pes cavus • Chronic osteomyelitis	• Whole spine MRI: diffuse mild small size of the entired spinal cord • Edx: diffuse motor polyneuropathy, not perform sensory studies • Nerve biopsy: axonal loss with endoneural fibrosis	***IGHMBP2*** (CMT2S/ 616155)	AR	-	-	• Autonomic involvement	-	Yes
CMT14	F	7M	• Foot drop • Distal weakness (LE>UE) • Distal paresthesia (LE>UE) • Equinovarus • Wheelchair-dependent since 6Y	• Muscle biopsy: neurogenic change • Edx: diffuse axonal sensorimotor polyneuropathy • *PMP22* dup: negative	***IGHMBP2*** (CMT2S/ 616155)	AR	-	-	• Autonomic involvement	• Family testing (father, mother)	Yes
CMT15	F	3Y	• Abnormal gait • Distal weakness (LE>UE) • Distal paresthesia (LE)	• Edx: diffuse axonal sensorimotor polyneuropathy • *PMP22* dup: negative	***MFN2*** (CMT2A2A/ 609260)	AD	-	-	• Optic atrophy	• Family testing (father)	Yes
CMT16	F	3Y	• Distal weakness (LE>UE) • Pes cavus	• Edx: diffuse axonal sensorimotor polyneuropathy • *PMP22* dup: negative	***MFN2*** (CMT2A2A/ 609260)	AD	-	-	• Optic atrophy	-	Yes

CMT17	F	1Y	• Foot drop • Distal weakness (LE>UE) • Bilateral optic atrophy (13Y) • Wheelchair-dependent since 18Y	• Edx: diffuse sensorimotor polyneuropathy (undetermined type) • *PMP22* dup: negative • Brain MRI: diffuse cerebellar atrophy, Atrophic change of bilateral optic nerves, optic chiasm and optic tracts.	***MFN2*** (CMT2A2A/ 609260) (CMT2A2B/ 617087)	AR	-	-	• Optic atrophy	• Family testing (father, mother)	Yes
CMT18	M	1Y	• Delay walking, tiptoe walking • Distal weakness (LE>UE)	• CK 95 U/L • Muscle biopsy: chronic denervation with re-innervation, marked endoneural fibrosis with decreased myelinated fibers • PCR-RFLP for *SMN1*: negative • Edx: diffuse sensorimotor polyneuropathy (undetermined type) • *PMP22* dup: negative	***MFN2*** (CMT2A2A/ 609260)	AD	-	-	• Optic atrophy	• Family testing (father, mother)	Yes
CMT19	M	1Y9M	• Frequent falling • Distal weakness (LE>UE) • Distal paresthesia (LE>UE)	• Edx: diffuse axonal sensorimotor polyneuropathy • *PMP22* dup: negative	***MFN2*** (CMT2A2A/ 609260)	AD	-	-	• Optic atrophy	• Family testing (father, mother)	Yes
CMT20	M	2Y	• Unable to jump • Distal weakness (LE>UE) • Distal paresthesia (LE)	• CK 89 U/L • Edx: diffuse axonal sensorimotor polyneuropathy • *PMP22* dup: negative	***MFN2*** (CMT2A2A/ 609260)	AD	-	-	• Optic atrophy	• Family testing (father, mother)	Yes
CMT21	F	2Y	• Walking difficulty • Distal weakness (LE, UE) • Pes planus	• Edx: diffuse axonal sensorimotor polyneuropathy	***MFN2*** (CMT2A2A/ 609260)	AD	-	-	• Optic atrophy	-	Yes
CMT22	F	1Y3M	• Abnormal gait • Distal weakness (LE, UE) • Distal paresthesia (LE, UE) • Pes planus, engrifted hand • Rhomberg sign	• Edx: diffuse axonal sensorimotor polyneuropathy • PMP22 dup: negative	***MFN2*** (CMT2A2A/ 609260)	AD	-	-	• Optic atrophy	• Family testing (father, mother)	Yes
CMT23	M	2Y	• Equinovarus, pes cavus, claw toes • Distal weakness (LE) • Distal muscle atrophy (LE)	• Edx: diffuse sensorimotor polyneuropathy (undetermined type) • PMP22 dup: negative	***MFN2*** (CMT2A2A/ 609260)	AD	-	-	• Optic atrophy	• Family testing (father, mother)	Yes
CMT24	F	8Y	• Calf pain • Distal weakness (LE, UE) • Distal paresthesia (LE, UE)	• Edx: diffuse axonal sensorimotor polyneuropathy • PMP22 dup: negative	***MFN2*** (CMT2A2A/ 609260)	AD	-	-	• Optic atrophy	-	Yes
CMT25	F	1Y3M	• Delay walking • GDD with articulation disorder • Distal weakness (LE, UE) • Genu recurvatum	• CK 207 U/L • PCR-RFLP for SMN1: negative • Edx: diffuse demyelinating sensorimotor polyneuropathy • PMP22 dup: negative	***MPZ*** (CMT1B/ 118200)	AD	-	-	• Hearing loss	• Family testing (father, mother)	Yes
CMT26	M	1Y	• Congenital severe scoliosis • Delay sitting • Distal weakness (LE>UE) • Genu recurvatum • Sleep breathing disorder on BiPAP	• MLPA for SMN1: negative • Edx: diffuse demyelinating sensorimotor polyneuropathy • PMP22 dup: negative	***MPZ*** (CMT1B/ 118200)	AD	-	-	• Hearing loss	• Family testing (mother)	Yes

CMT27	M	4Y	• Delay walking • Distal weakness (LE>UE) • Scoliosis	• Edx: diffuse demyelinating sensorimotor polyneuropathy • PMP22 dup: negative	***MPZ*** (CMT1B/ 118200)	AD	-	-	• Hearing loss	• Family testing (father, mother)	Yes
CMT28	M	12Y	• Frequent falling • Distal weakness (LE>UE) • Bilateral SNHL	• Edx: diffuse demyelinating sensorimotor polyneuropathy • PMP22 dup: negative	***NEFL*** (CMT1F/ 607734)	AD	-	-	• Hearing loss	• Family testing (father, mother)	-
CMT29	M	2Y	• Walking difficulty • Distal weakness (LE, UE) • Distal paresthesia (LE, UE) • Pes cavus, hammer toe, claw hand • Bilateral SNHL • Neurogenic bladder (6Y)	• CK 271 U/L • Edx: diffuse demyelinating sensorimotor polyneuropathy • PMP22 dup: negative	***NEFL*** (CMT1F/ 607734)	AD	-	-	• Hearing loss	• Family testing (father, mother)	-
CMT30	M	1Y3M	• Walking difficulty • Distal weakness (LE, UE) • Distal paresthesia (LE) • Pes cavus • Left mild SNHL	• Edx: diffuse demyelinating sensorimotor polyneuropathy • PMP22 dup: negative	***NEFL*** (CMT1F/ 607734)	AD	-	-	• Hearing loss	• Family testing (father, mother)	-
CMT31	M	3Y	• Distal weakness (LE) • Distal paresthesia (LE) • Rhomberg sign	• Edx: diffuse demyelinating sensorimotor polyneuropathy • PMP22 dup: negative	***PMP2*** (CMT1G/ 618279)	AD	-	-	-	• Family testing (mother)	Yes
CMT32	M	1Y	• Delay walking and unsteady gait • Distal weakness (LE, UE) • Distal paresthesia (LE, UE) • Ataxia	• Brain MRI: NL • Edx: diffuse sensorimotor polyneuropathy (undetermined type) • PMP22 dup: negative	***PMP22*** (CMT1E/ 118300)	AD	-	-	• Hearing loss	• Family testing (father, mother)	-
CMT33	M	1Y3M	• Delay walking • Distal weakness (LE, UE) • Distal paresthesia (LE, UE)	• Edx: diffuse sensorimotor polyneuropathy (undetermined type) • PMP22 dup: negative	***PMP22*** (CMT1E/ 118300)	AD	-	-	• Hearing loss	-	-
CMT34	F	2Y	• Walking difficulty • Distal weakness (LE, UE) • Pes cavus	• Edx: diffuse demyelinating sensorimotor polyneuropathy	***SH3TC2*** (CMT4C/ 601596)	AR	-	-	• Hearing loss • Scoliosis	-	Yes
HSN1	M	11Y	• Foot drop • Distal weakness (LE>UE) • Distal paresthesia (LE, UE)	• Edx: diffuse axonal sensorimotor polyneuropathy (sensory>motor)	***KIF1A*** (HSN2C/ 614213)	AR	-	-	-	-	-
HSN2	F	7M	• Tongue biting and self-multilation • Constipation and bloody diarrhea • Corneal ulcers (neuopathic keratitis) • Severe Intellectual disability	• Edx: axonal predominate sensory polyneuropathy	***PRDM12*** (HSAN8/ 616488)	AR	-	-	-	• Family testing	-
Group 3: Congenital myasthenic syndrome											
CMS1	F	6M	• Bilateral ptosis • Hypotonia • Waddling gait	• CK 37 U/L • Anti-AchR positive	***CHRNE*** (CMS4A/ 605809)	AD	Avoid muscle biopsy	• Discontinue AchEI (previously treated as ocular MG) • Fluoxetine	-	• Family testing (father, mother)	Yes
CMS2	F	2W	• Bilateral ptosis with opthalmoplegia • Recurrent respiratory failure	• CK 45 U/L, lactate 1.3 mmol/L • Muscle biopsy: no specific change • Edx: NL, RNS: decremental response of CMAP at 2-3 Hz stimulation	***COLQ*** (CMS5/ 603034)	AR	-	• Discontinue AchEI (previously treated as MG) • Salbutamol, ephedrine	-	• Family testing (father, mother)	Yes

CMS3	F	birth	• Bilateral ptosis with opthalmoplegia • Severe laryngomalacia • Recurrent respiratory failure	• CK 22 U/L, lactate 1.2 mmol/L • Anti-AchR negative • Edx: NL, RNS: NL • Muscle biopsy: non-specific change, no definite structural anomaly seen	***COLQ*** (CMS5/ 603034)	AR	-	• Avoid AchEI • Salbutamol, ephedrine	-	• Family testing (father, mother)	Yes
CMS4	M	birth	• Bilateral ptosis with opthalmoplegia • Marked hypotonia • Severe scoliosis with excessive lumbar lordosis • Recurrent respiratory failure	• CK 39 U/L • Anti-AchR negative • Edx: NL, RNS: decremental response of CMAP at 2-3 Hz stimulation • Muscle biopsy: no specific change	***COLQ*** (CMS5/ 603034)	AR	-	• Avoid AchEI • Salbutamol, ephedrine	-	-	Yes
CMS5	M	1Y	• GDD, FTT • Hypotonia • Recurrent pneumonia • Bilateral ptosis with opthalmoplegia • Gowers' sign • Mild scoliosis with lordosis	• Plasma amino acid, urine organic acid: NL, NH_3 35 umol/L • CK 96 U/L, lacate 2.4 mmol/L • Brain MRI and MRS: NL • Muscle biopsy: NL • Edx: NL, RNS: decremental response of CMAP at 3 Hz stimulation	***COLQ*** (CMS5/ 603034)	AR	-	• Avoid AchEI • Salbutamol, ephedrine	-	-	Yes
CMS6	M	10Y	• Proximal muscle weakness	• CK 159 U/L • Edx: chronic myopathic change, RNS: decremental response of CMAP at 2 Hz stimulation • Muscle biopsy: mild myopathic change, non-specific	***COLQ*** (CMS5/ 603034)	AR	-	• Avoid AchEI • Salbutamol, ephedrine	• Respiratory involvement	-	Yes
CMS7	M	1M	• Bilateral ptosis with opthalmoplegia • Proximal muscle weakness • Scoliosis	• Edx: chronic myopathic change, RNS: decremental response of CMAP at 3 Hz stimulation with repetitive CMAP	***COLQ*** (CMS5/ 603034)	AR	Avoid muscle biopsy	• Avoid AchEI • Salbutamol, ephedrine	• Respiratory involvement	-	Yes
CMS8*	M	19Y	• Bilateral ptosis • Facial weakness • Proximal muscle weakness	• Anti-AchR negative • Muscle biopsy: mild myopathic change • Edx: chronic myopathic change, RNS: decremental response of CMAP at 3 Hz stimulation	***RAPSN*** (CMS11/ 616326)	AR	-	• AchEI	• Respiratory involvement	• Family testing (sister)	Yes
CMS9*	F	5Y	• Bilateral ptosis • Facial and bulbar weakness • Proximal muscle weakness	• Anti-AchR negative • RNS: decremental response of CMAP at 3 Hz stimulation	***RAPSN*** (CMS11/ 616326)	AR	-	• AchEI	• Respiratory involvement	• Family testing (brother)	Yes
CMS10	M	birth	• Congenital hypotonia • Mild ptosis with ophthalmoplegia • Facial and bulbar weakness Proximak muscle weakness	• CK: NL • Anti-AchR: negative • RNS: decremental response of CMAP at 3 Hz stimulation	***RAPSN*** (CMS11/ 616326)	AR	Avoid muscle biopsy	• AchEI	• Respiratory involvement	• Family testing (father, mother)	Yes
Group 4: Motor neuron disease											
MND1	F	9M	• Hypotonia • Myopathic face • Proximal muscle weakness • Areflexia • Bilateral diaphragmatic paralysis • Myoclonic epilepsy (develop after genetic diagnosis)	• CK 1475 U/L • Muscle biopsy: neurogenic change • Brain MRI: NL • MLPA for *SMN1*: negative	***ASAH1*** (Spinal muscular atrophy with progressive myoclonic epilepsy/ 159950)	AR	-	• ASM • Tracheostomy	• SNHL • Epilepsy • Scoliosis	• Family testing (father, mother, brother) • Early diagnosis of her affected brother	Yes

Abbreviations: AAO, age at onset; ACEI, angiotensin-converting enzyme inhibitor; AchEI, acetylcholine esterase inhibitor; AD, autosomal dominant; ADHD, attention deficit hyperactive disorder; AMC, arthrogryposis multiplex congenita; AR, autosomal recessive; ARB, angiotensin receptor blocker; BiPAP, bilevel positive airway pressure; CIDP, chronic inflammatory demyelinating polyradiculoneuropathy; CK, creatine kinase (normal: < 190 U/L); CM, congenital myopathy; CMAP, compound muscle

action potential; CMS, congenital myasthenic syndrome; CN, cranial nerve; COL6, collagen 6; CV, conduction velocity; DMD, Duchenne muscular dystrophy; ECG, electrocardiogram; Edx, electrodiagnostic testing; EEG, electroencephalogram; EF, ejection fraction (from echocardiogram); F, female; GA, gestational age; GDD, global developmental delay; LE, lower extremities; LVNC, left ventricular noncompaction; M, male; MD, muscular dystrophy; MLPA, multiplex ligation-dependent probe amplification; MOI, mode of inheritance; MRI, magnetic resonance imaging; MRS, magnetic resonance spectroscopy; NIV, noninvasive ventilation; NL, normal; OSA, obstructive sleep apnea; PCR-RFLP, polymerase chain reaction-restriction fragment length polymorphism; PFT, pulmonary function test; RNS, repetitive nerve stimulation; SNHL, sensoineural hearing loss; SSCD, sarcolemma specific collagen deficiency; UE, upper extremities; XD, X-linked dominant; XR, X-linked recessive

* sibling relationship

In the patient-ID column, CM, DMD, MD, MM, MC, CMT, HSN, CMS, and MND represent patients clinically diagnosed with congenital myopathies, Duchenne muscular dystrophy, other muscular dystrophies, metabolic/mitochndrial myopathies, muscle channelopathy, Charcot–Marie–Tooth disease, hereditary sensory neuropathy, congenital myasthenic syndrome, and motor neuron disease, respectively.

In the AAO column, D, M, W, and Y represent day(s), month(s), week(s), and year(s), respectively.

In the conventional investigation column, Edx with CV of < 35 m/s in the upper extremity nerve indicated the demyelinating type, a CV of 35–45 m/s was classified as intermediate, and a CV of > 45 m/s was considered axonal.