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REVIEW ARTICLE

Orthopaedic abnormalities in primary myopathies

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Orthopaedic abnormalities are frequently recognised in patients with myopathy but are hardly systematically reviewed with regard to type of myopathy, type of orthopaedic problem, and orthopaedic management. This review aims to summarize recent findings and current knowledge about orthopaedic abnormalities in these patients, their frequency, and possible therapeutic interventions.

A MEDLINE search for the combination of specific terms was carried out and appropriate articles were reviewed for the type of myopathy, types of orthopaedic abnormalities, frequency of orthopaedic abnormalities, and possible therapeutic interventions.

Orthopaedic abnormalities in myopathies can be most simply classified according to the anatomical location into those of : the spine, including dropped head, camptocormia, scoliosis, hyperlordosis, hyperkyphosis, or rigid spine ; the thorax, including pectus excavatum (cobbler's chest), anterior/posterior flattening, or pectus carinatum (pigeon's chest) ; the limb girdles, including scapular winging and pelvic deformities ; and the extremities, including contractures, hyperlaxity of joints, and hand or foot deformities. These orthopaedic abnormalities can be most frequently found in arthrogryposis, muscular dystrophies, congenital myopathies, myofibrillar myopathies, and myotonic dystrophies. Occasionally, they also occur in metabolic myopathies or other types of myopathy. Most of the orthopaedic abnormalities are sufficiently accessible to conservative or surgical orthopaedic treatment.

Orthopaedic abnormalities have major implications in the management and outcome of myopathy patients ; they should be closely monitored and treated on time.

Keywords : myopathy ; muscular dystrophy ; neuromuscular disorder ; orthopaedic disorders ; surgery.

LIST OF ABBREVIATIONS

AMC	Arthrogryposis multiplex congenita
BMD	Becker muscular dystrophy
CCD	Central core disease
CMD	Congenital muscular dystrophy
CMP	Congenital myopathy
DHS	Dropped head syndrome
DMD	Duchenne muscular dystrophy
EDMD	Emery-Dreifuss muscular dystrophy
FHL1	Four-and-a-half **L**in11, **I**sl-1, **M**ec-3-domain 1 gene
FSH	Facioscapulohumeral muscular dystrophy
LGMD	Limb girdle muscular dystrophy
LMNA	Lamin A/C
MD1	Myotonic dystrophy 1
MD	Muscular dystrophy
MP	Myopathy
MYH	Myosin heavy chain
NMDs	Neuromuscular disorders
PVCR	Posterior vertebral column resection

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RYR1	Ryanodine receptor 1
SEPN1	Selenoprotein 1
TPM	Tropomyosin
USS	Universal spine system
VC	Vital capacity
VCP	Valosin-containing protein

INTRODUCTION

Though myopathies are per definition disorders exclusively affecting the skeletal muscles, they frequently cause secondary problems in other organs or systems as well. This is particularly the case for the cerebrum and the heart. In addition to these organs patients with myopathy frequently develop orthopaedic abnormalities and in single cases, such as arthrogryposis multiplex congenita (AMC, multiple joint contractures and talipes present at birth) or Escobar syndrome (multiple pterygium syndrome), the orthopaedic abnormality dominates the phenotype (Table I) (*84,116*). Since orthopaedic problems can substantially influence daily living, they are a major issue in the management of these patients. The following review aims to summarize the current knowledge and recent findings about orthopaedic abnormalities in patients with myopathies, their frequency in these patients and frequently utilized therapeutic interventions.

METHODS

A MEDLINE search for the terms "spinal deformity", "dropped head", "camptocormia", "bent spine", "hyperlordosis", "hyperkyphosis", "rigid spine", "thoracic deformity", "pectus excavatum" ("cobbler's chest"), "pectus carinatum" ("pigeon's chest"), "scapula alata", "scapular winging", "winging scapula", "scapulopexy" (fixation of the scapula to the thorax), "scapulothoracic arthrodesis" (fixation of the shoulder blade to the thoracic wall), "pelvic obliquity", "pelvic tilt", "windblown hip", "windswept hip", "contracture", "contractures", "genu recurvatum", "genu valgus", "genu varus", "tip-toe walking", "joint laxity", "pes varus", "pes valgus", "pes cavus", "pes equinovarus", "talipes", "pes planovalgus", "Achilles tendon contracture", and "arthrogryposis" in combination with "myopathy", "myopathies", "neuromuscular disorder", and "skeletal muscle" was carried out and appropriate articles reviewed for the type of myopathy, types of orthopaedic problems, frequency of orthopaedic problems, and possible therapeutic options in these patients.

Table I. — Orthopaedic problems in myopathies

Spine
– Dropped head syndrome
– Bent-spine syndrome (camptocormia)
– Scoliosis
– Hyperlordosis
– Hyperkyphosis (gibbus)
– Rigid spine
Shoulder girdle
– Scapula alata
Thorax (chest)
– Cobbler's chest (pectus excavatum)
– Anterior/posterior flattening of chest cavity
– Pectus carinatum (pigeon's chest)
Pelvic deformities
– Anterior/posterior tilt
– Pelvic obliquity
– Windblown (windswept) hip
Extremities
– Contractures of distal / proximal joints, small / large joints, spine (rigid spine)
– Joint hypermotility / hyperlaxity
– Luxation of joints, hips
– Hand / foot deformities
– Tip-toe, pes equinovarus (clubfoot, talipes), pes planovalgus

RESULTS

Reviewing the papers selected according to the criteria mentioned above, it turned out that a number of different orthopaedic abnormalities occur in patients with myopathies. Generally, a mobility unit, such as a joint or the intervertebral discs, may undergo pathological alterations, which lead to complete immobility of the unit, decreased motility, or hypermotility. These changes most frequently include irregular positioning or dislocation of the combined elements. Rarely, there may be deformities, which are not accompanied by impaired motility of the affected joints. Accordingly, orthopaedic abnormalities in myopathies include : spinal abnormalities, such as dropped head syndrome (DHS), camptocormia, scoliosis, hyperlordosis, hyperkyphosis, or rigid spine ; thoracic deformities or

impaired mobility of the thorax, such as pectus excavatum, pectus carinatum ; shoulder girdle or pelvic girdle abnormalities, such as scapular winging (scapula alata), anterior / posterior pelvic tilt, pelvic obliquity or windblown (windswept) hip ; limb abnormalities, such as contractures of small or large joints with or without deformities of the limbs, hypermobility or hyperlaxity of joints with or without habitual subluxation or luxation of joints, or hand or foot deformities (Table I). Most deformities of the extremities result from a combination of muscle weakness and imbalance, and surgical procedures aim at correcting these deformities and rebalancing existing musculature by release or transfer (*4*).

Spine

Dropped head syndrome

DHS, also known as cervical kyphosis or floppy head syndrome, is characterized by the inability of the patient to voluntarily lift his head from the chin-on-chest position into the upright position due to weakness of the head extensor muscles. Weakness of these muscles may be complete or incomplete, allowing at least a limited head lift. Dropping of the head may either go along with neck pain or may be painless. DHS was initially described in myasthenia gravis and motor neuron disease. Before the head drop can be attributed to a primary muscle disease, a number of differentials, including secondary muscle disorders and central nervous system (CNS) disease have to be excluded. Secondary muscle disorders, which need to be considered in paediatric and adult patients include focal myositis (idiopathic inflammatory myopathy), polymyositis, dermatomyositis, hypothyroidism, hyperparathyroidism, Cushing syndrome, secondary carnitine deficiency, syringomyelia, and neuromuscular transmission defects. Differential diagnoses predominantly occurring in adults include Parkinsonism, multisystem atrophy, Lewy body disease, basal ganglia disease, inclusion body myositis, liquorice-induced hypokalemia, drugs, such as olanzapine or chemotherapeutics, radiotherapy, or paraneoplasia. Primary myopathies, in which DHS has been described so far are listed in table II. Therapeutic approaches for DHS in primary myopathies include physiotherapy, neck extension orthoses, analgetics, or surgical stabilisation of the cervical spine in selected cases. The latter may be carried out by instrumented, posterior, cervicothoracic spinal fusion (*37*).

Table II. — Myopathies associated with dropped head syndrome

Muscular dystrophies
– FSH*
– Laminopathy
– CMD (LMNA)
Congenital myopathies
– Nemaline myopathy
– Congenital myopathy
Myotonic myopathies
– Myotonic dystrophy type 1#
– Myotonic dystrophy type 2#
Metabolic myopathies
– Mitochondrial myopathy*
– Acid maltase deficiency (Pompe)
Unclassified myopathies
– Isolated cervical myopathy

* : variable childhood or adult onset, # : predominantly adult onset.

Bent spine syndrome (camptocormia)

Camptocormia, also known as bent spine syndrome or progressive lumbar kyphosis, is an acquired postural abnormality with forward-flexed posture, leading to prominent lumbar kyphosis. Camptocormia usually increases when the patient gets up or walks (*5,21*). Occasionally, camptocormia is associated with low back pain, and weakness of the gluteus maximus, hip, or genu flexion (*5*) but can be relieved in recumbent or supine position. The diagnosis may be established upon clinical findings, electromyography, or muscle biopsy, which may be normal or may show mild myopathic features, inflammatory features suggesting focal inflammatory myopathy (focal myositis), or dystrophic features. Myopathies, in which bent spine syndrome has been described so far include dysferlinopathy (*124*) or nemaline myopathy (*100*) in paediatric and adult patients and myotonic

dystrophy type 1 (*25,60*), myotonic dystrophy type 2 (*125*), or axial myopathy (*130*) in adult patients. Camptocormia, however, occurs together with a number of other conditions, which have to be excluded before attributing the abnormality to a primary myopathy. Non-myopathic conditions associated with camptocormia include dermatomyositis, polymyositis, trauma, and malignancies in paediatric and adult patients and dystonia, multisystem atrophy, Tourette syndrome, conversion disorder, hypothyroid myopathy, inclusion body myositis, chronic axial myositis (*91*), myasthenia gravis, amyotrophic lateral sclerosis, chronic inflammatory demyelinating polyneuropathy, lumbar disc herniation, and occasionally, medication with olanzapine or valproate in adult patients. Camptocormia is treated according to the underlying disorder. If camptocormia is due to Parkinson's disease, dystonia or multi-system atrophy, administration of L-DOPA can be helpful. In single cases, pallidal high-frequency deep brain stimulation or bilateral subthalamic nucleus stimulation may have a beneficial effect (*35*). If camptocormia is induced by olanzapine, electroconvulsive therapy may be effective. Injection of botulinum toxin into the rectus abdominis muscles may also relieve the abnormal posture. If conservative measures are ineffective surgical posterior thoracolumbar fixation with anterior body fusion may be an alternative (*103*).

Scoliosis

Scoliosis is a frequent and often dominant feature of neuromuscular disorders (NMDs). Scoliosis is defined as twisting and torque of spinal column in all axes. Usually, scoliosis presents with a double S-shape in the frontal plane. Scoliosis can be quantified by measuring various angles. The most well known and most widely used is Cobb's angle (angle between two lines drawn perpendicular to a line along the superior endplate of the superior end vertebra and a second line drawn along the inferior endplate of the inferior end vertebra). Primary myopathies, in which scoliosis has been found so far are listed in table III. Scoliosis can be most frequently found in muscular dystrophies and congenital myopathies (CMPs) (Table III). Treatment of scoliosis is based either on conservative or surgical measures of correction with advantages and disadvantages of either method. Which patients profit more from the one or the other approach is under debate. Conservative measures include physiotherapy, plaster casts, and bodices. Disadvantages of plaster casts are that the patient is somehow immobilized for spinal movements and that thoracic expansion might be restricted during inspiration.

There are a number of different surgical approaches to help the patients. Generally, surgical scoliosis correction may be achieved by a posterior approach, by an anterior approach, or by a combined anterior posterior surgical approach (*137*). The most frequent posterior only approaches for scoliosis correction are pedicle screw fixation, posterior spinal fusion, or segmental spinal instrumentation (*82*). To avoid an anterior approach, apical axial derotation of neuromuscular scoliosis can be sufficiently achieved by a posterior only pedicle screw fixation (*77,82*). Such an approach will improve sitting and balance, and will also reduce Cobb's angle, pelvic obliquity, and apical rotation (*82*). Another technique for scoliosis stabilization is dorsal spondylodesis with universal spine system (USS) instrumentation (*104*). Since anterior procedures might compromise pulmonary function and since posterior vertebral column resection (PVCR) might carry the risk of neurologic injuries, posterior multilevel vertebral osteotomy was recently proposed as an alternative for correction of severe and rigid neuromuscular scoliosis (*137*). With such an approach Cobb's angle, pelvic obliquity, and apical rotation can be significantly reduced (*137*). Pelvic fixation in scoliosis surgery is usually carried out with iliac screws similar to the Galveston technique (*44*). Pedicle screw fixation results in reduction of the Cobb angle, pelvic tilt, and lumbar lordosis (*44*). Thoracic kyphosis usually remains unchanged (*44*). Instead of Cobb's angle some authors propose a vital capacity below 30% as an indicator for scoliosis surgery. If a myopathic condition is suspected as the underlying cause of scoliosis, biopsy of the paraspinal muscles during scoliosis surgery particularly in the absence of limb muscle weakness, is recommended. In such cases

Table III. — Myopathies associated with scoliosis

Myopathy	Reference
Muscular dystrophies	
– Dystrophinopathy (in 90% of the cases)	(22,44,54,58,81)
– FSH*	(151)
– Laminopathy (LMNA)	(110,106,147)
– Ullrich CMD	(67)
– Bethlem CMD	(79)
– Rigid spine syndrome	(135)
Congenital myopathies	
– Congenital fiber type disproportion (SEPN1)	(17,18)
– CMP with type 1 fiber predominance	(88)
– Multi-minicore disease	(119)
– CCD	(107,126)
– Multicore disease	(104)
– CMP	(15)
– Escobar syndrome with nemaline MP (TPM2)	(23)
– Reducing body myopathy (FHL1)	(122,127)
– Arthrogryposis	(64)
Distal myopathies	
– Distal myopathy with normal dysferlin	(87)
Myofibrillar myopathy*	(personal communication)
Myotonic syndromes	
– MD1#	(141)
– Congenital MD1	(11)
Other	
– Native American myopathy	(132)

* : variable childhood or adult onset, # : adult onset.

muscle biopsy may help to establish the diagnosis, particularly if congenital myopathy is present.

Hyperlordosis

Hyperlordosis of the spine may either concern the lumbar or cervical spine, whereby lumbar hyperlordosis is much more frequent than cervical hyperlordosis. In the majority of the cases hyperlordosis is associated with other orthopaedic problems (*93*), such as pelvic deformity or scoliosis, particularly if the lumbar spine is affected. Primary myopathies, in which hyperlordosis has been described as a phenotypic feature include the dystrophinopathies, some of the limb girdle muscular dystrophies (LGMDs), and some congenital muscular dystrophies (CMDs) in paediatric patients, and facio-scapulo-humeral (FSH) muscular dystrophy and laminopathies in adult patients (Table IV). There are only limited therapeutic options for hyperlordosis of the cervical spine. Neck hyperextension in Duchenne muscular dystrophy (DMD) may be surgically treated with posterior interspinous fusion (*38*). The procedure consists of a posterior approach to the cervical spine and correction of hyperextension by releasing fibrotic muscles and ligaments and stabilization of bone grafts driven into the interspinous spaces (*38*). Correction of cervical hyperextension may be also carried out by posterior release with a halo extension, but the perioperative risk appears to be high (*105*). Surgical treatment of lumbar hyperlordosis is similar to that of scoliosis and usually combined with pelvic fixation with iliac screws similar to the Galveston technique (*44*).

Hyperkyphosis (gibbus)

Hyperkyphosis of the thoracic spine, also known as severe cervico-thoracic kyphosis or gibbus, may

Table IV. — Myopathies associated with hyperlordosis, hyperkyphosis, or rigid spine

Myopathy	Hyperlordosis	Hyperkyphosis	Rigidity	Reference
DMD (dystrophin)	y	y	nr	(38)
LGMD2A (calpain)*	y	nr	nr	(73)
LGMD2C (g-sarcogylcan)	y	nr	nr	(80)
LGMD2G (telethonin)	y	nr	nr	(97)
Laminopathy (LMNA)	y	y	y	(106,157)
FSH*	y	nr	nr	(24,57,59,58)
X-EDMD (Emerin)	nr	nr	y	(6)
EDMD	nr	nr	y	(41)
Rigid spine syndrome (FHL1)	nr	y	y	(123,135)
Rigid spine (SEPN1)	nr	nr	y	(140)
Rigid spine (dysferlin)	nr	nr	y	(72,90,127)
Ullrich CMD (COLA1-3)	nr	nr	y	(8)
Scapuloperoneal myopathy (FHL1)#	nr	nr	y	(123)
Central core disease	y	nr	nr	(69)
CMP with type 1 predominance	nr	y	nr	(88)
CMP with tubular aggregates	y	nr	nr	(28)
Multicore disease	y	nr	y	(104,144)
Multiminicore disease (SEPN1)	nr	nr	y	(158)
Reducing body myopathy (FHL1)	nr	nr	y	(27,94,122)
Congenital myotonic dystrophy	y	y	nr	(55,10)
Mitochondrial depletion syndrome (TK2)	nr	nr	y	(99)
Acid maltase deficiency	nr	nr	y	(61)
Native American myopathy	nr	y	nr	(132)

Nr : not reported, * : variable childhood or adult onset, # : adult onset.

be flexible or fixed and is characterized by increased bending of the thoracic spine in the sagittal plane. As with other spinal deformities, hyperkyphosis usually does not occur isolated but in the majority of the cases in combination with other orthopaedic problems, most frequently with scoliosis or hyperlordosis. Primary myopathies with hyperkyphosis include myotonic dystrophy type 1 (MD1) (*55*), DMD (*49,102*), actin-myopathy (ACTA1) (*40*), or axial myopathy (*130*). Correction of hyperkyphosis may be carried out by straightening of the deformed spinal segment by rod and screw instrumentation to establish an arthrodesis (*37*). Stabilisation may be carried out as a two-stage procedure with initial discectomies and fusions and secondary Smith-Petersen osteotomy with pedicle screw fixation, which may cause neurological side effects (*55*).

Rigid spine (limit spine)

Rigid spine is characterized by partial or complete restriction of two, some, or all vertebral segments to move against each other. In severe forms the patient is unable to move his head in any direction and to bend or rotate his spine. The cause of the rigidity may be multifactorial due to contractures of the intervertebral joints, rigidity or stiffness of the axial spinal musculature, calcification of ligaments, or blocking between two or several neighbouring vertebrae. Rigid spine is the dominant feature in a CMD termed rigid spine syndrome, which is due to

mutations in the SEPN1 gene but has been described in a number of other myopathic conditions as well (Table IV). Treatment of choice is physiotherapy but in an increasing number of mostly severe cases with rigid curves surgical measures are proposed. The most well known is posterior multilevel, vertebral osteotomy, which has the advantage that it provides also an anterior column release without an anterior approach (*137*). Other spinal abnormalities in myopathies include block vertebrae, vertebral fusion, short neck, fused ribs or spina bifida occulta as has been described in a patient with central core disease (CCD) due to a RYR1 mutation (*120*).

Shoulder girdle

Scapula alata

The most prominent orthopaedic abnormality on the shoulder girdle is the loose scapula, also known as scapular winging, winging scapula, or scapula alata. Scapular winging is characterized by spontaneous or provoked unilateral or bilateral protrusion of the shoulder blade from the underlying dorsal thoracic wall. If winging of the scapula is absent at rest or only hardly visible it may be provoked or enhanced by the following maneuvers : abduction of the stretched arms against the resistance of the investigator ; pressing together both hand palms in front of the chest ; outward rotation of the lower arms in the shoulders ; or adduction of the bent elbows at the back against the investigator's resistance. There are a number of differential diagnoses, which have to be thoroughly ruled out before winging of the scapula can be definitively attributed to an underlying myopathy. These include radiculopathy, plexopathy, or lesions of the long thoracic nerve. Myopathies, in which winging of the scapula is a dominant feature are the FSH, where winging of the scapula is an obligate aspect of the phenotype, LGMDs, such as laminopathy, LGMD2A (*42,73*), LGMD2C, LGMD2D (*31*), LGMD2G (*97*), or unclassified LGMD (*26*), CMDs, some CMPs, or Pompe's disease (Table V). There is no therapeutic algorithm for scapula alata (*2*), but if proximal functions of the upper extremity are impaired to such a degree that the patient is handicapped, fixation of the shoulder blade to the thoracic wall is indicated. Techniques most frequently applied for this purpose are scapulothoracic arthrodesis or interscapulo-scapulocostal scapulopexy (*46,65,114*).

Thorax

Chest deformities are a frequent orthopaedic problem in primary myopathies and occur most frequently in combination with other orthopaedic abnormalities, most frequently with scoliosis. Thoracic deformities are often a direct consequence of the spinal problem and may be classified as anterior or posterior chest wall deformities. If untreated, thoracic deformities have a strong impact on the prognosis of the individual since they can strongly impair respiration and cardiac function. If the thoracic cavity can't sufficiently expand during inspiration restrictive pulmonary disease develops. Additionally, heart failure may develop if relaxation during diastole and thus filling of the left ventricle is restricted. Anterior chest or thoracic deformities associated with myopathies include pectus excavatum, also known as cobbler's chest, anterior / posterior flattening of the chest cavity, and pectus carinatum (Table VI). More rare thoracic deformities include bell-shaped chest cavity in actinmyopathy (*40*) or fused ribs in CCD (*120*). Thoracic deformities require surgical correction if respiration and diastolic filling are impaired. Surgical corrections focus on the correction of pectus excavatum and pectus carinatum. Pectus excavatum is surgically corrected by a sternochondroplasty with bilateral subperichondrial resection of the deformed costal cartilages and sternal osteotomy resecting a wedge of the anterior cortex and fracturing the posterior cortex. Anterior displacement is maintained with silk sutures closing the osteotomy defect. The sternum may be secured by intramedullary fixation with a Steinman pin (*128*). Surgical correction of pectus excavatum may be also carried out by subperichondral resection of deformed costal cartilages, detachment of the xiphoid process, transverse sternotomy at the upper level of the deformed sternum, which is then bent forward, and by securing of

Tables V. — Myopathies associated with winging of the scapula

Disorder	Reference
FSH*	(26,114)
LGMD2A (calpain)*	(42,73)
LGMD2C (gamma-sarcoglycanopathy)	(80)
LGMD2D (alpha-sarcoglycanopathy)	(31,83)
LGMD2G (telethonin)	(97)
EDMD (LMNA)	(6)
Laminopathy	(106)
CMD	(internet)
Cap myopathy (CMP)	(20)
Valosin-containing protein (VCP) myopathy*	(134)
Actin-myopathy (ACTA1, congenital)	(40)
Acid-maltase deficiency (Pompe)	(26)

* : variable childhood or adult onset.

Table VI. — Myopathies with thoracic deformity, such as pectus excavatum (cobbler's chest, funnel chest), anterior/posterior flattening of chest cavity, pectus carinatum (pigeon's chest), or bell shaped chest

Myopathy	Reference
Duchenne muscular dystrophy	(153)
FSH*	(129)
Rigid spine syndrome	(135)
Non-Fukuyama CMD	(152)
Multi-minicore disease	(89)
Nemaline myopathy	(66,70)
Congenital myopathy	(15)
Multicore disease (thoracic deformity)	(104)
Myotubular (centronuclear) myopathy	(48,142)
Minimal change myopathy	(95)
Actin myopathy (congenital)	(40)
Arthrogryposis multiplex congenita	(118)
Distal arthrogryposis 5	(150)
Distal arthrogryposis (pectus excavatum)	(51)
Lipid raft disease	(53)

* : variable childhood or adult onset.

the corrected sternal position by a "hammock" of synthetic mesh, spread behind the sternum, and attached to the respective cartilage remnants (*115*). An alternative to sternochondroplasty is the minimal invasive Nuss procedure. If thoracic deformities result from scoliosis, stabilization of the spine is inevitable. Physiotherapy after surgery may support recovery from surgical intervention. The initial steps of pectus carinatum correction are similar to those of pectus excavatum correction but the sternum is not freed from its environment. A length of 3-4 cm is resected from the distal sternum and the

xyphoid process is reattached in the proper anatomical direction (*115*).

Pelvis

Pelvic deformities are a common orthopaedic problem in patients with NMDs and include excessive posterior or anterior tilt, pelvic obliquity, or windblown (windswept) hip syndrome (*113*). Posterior / anterior pelvic tilt is characterized by forward / backward tilting of the pelvis due to a spinal deformity, such as lumbar hyperlordosis or due to instability of the hips. Pelvic obliquity is characterized by tilting of the pelvis in the frontal plane to the right or left side. Tilting in the frontal plane may also derive from spinal deformity, from unilateral or uneven weakness of the pelvic girdle muscles, or from affection of the hips. Windblown deformity is defined by a malposition of the lower extremities, such that one leg is flexed in the hip, abducted and outwardly rotated, whereas the other leg is adducted, inwardly rotated and extended in the hip. Pelvic deformities always additionally affect the hips and result in contractures, subluxation, or dislocation (*10*). Pelvic obliquity in association with scoliosis may affect the sitting balance and may become painful (*10*). Pelvic deformities tend to cause subluxation or dislocation of the hips, usually on the high side of a tilted pelvis (*10*). Primary myopathies associated with pelvic deformity include DMD (*10,22*), laminopathy (*110*), LGMDs (*33*), or congenital MP with type 1 fiber predominance (*88*). Concerning the optimal therapy for various pelvic abnormalities, in particular hip disorder, the discussion is controversial (*10*). Surgical correction of a pelvic deformity is often only achievable if surgical correction of concomitant scoliosis is performed (*113*).

Extremities

Contractures

Contractures are the most frequent orthopaedic problem in patients with primary myopathies. Contractures may have a muscular (replacement of elastic muscle tissue by connective tissue) or articular cause and may result in complete (fixed) or incomplete restriction of mobility. Contractures may concern the distal or proximal joints, the small or large joints, one joint or several joints, the extremities or the spine. Contractures go along with deformities in the majority of the cases and may be associated with other orthopaedic abnormalities. In Ullrich CMD proximal contractures may go along with distal hyperlaxity in the same patient (*8*). In patients with primary myopathies the cause of joint contractures is more frequently due to the muscular problem than due to a primary joint problem. Considering the muscular causes it can be speculated to result from a mixture of immobility, imbalance between agonists and antagonists, impaired reciprocal inhibition, or changes between muscular hypertonia and hypotonia. Considering the articular causes it may be due to a cartilage abnormality or due to a primary articular process associated with the underlying NMD. Myopathies, which present with contractures are listed in Table VII. Joints most frequently affected in myopathies are the elbows and the hips. More rarely affected are the wrists, the knees and the small finger joints. The development of finger flexion contractures is a hallmark of Bethlem myopathy (*67*). These contractures have a dynamic nature in childhood, appearing and disappearing in various joints (*67*). Nearly all older patients develop fixed contractures of the finger flexors, wrists, elbows and ankles (*67*). If contractures predominate among the clinical manifestations, Bethlem myopathy resembles Emery-Dreifuss muscular dystrophy (EDMD) (*67*). Contractures of all or the distal joints are also a hallmark of the various types of arthrogryposis, CMPs with prominent orthopaedic abnormalities due to mutations in proteins of the contractile apparatus, such as troponin, tropomyosin, cofilin, or MYH3 (Table VII) (*43*). If the knee is affected contractures lead to hyperextension (*7*). The most favored way to manage contractures is prophylaxis by preserving mobility by physiotherapy as well as canes, braces, orthoses, splints, or plaster casts. In a study on 144 DMD patients, contractures were managed by stretching exercises, prescribed periods of standing and walking, or application of knee-ankle foot orthoses with a temporary effect (*46*). In a

Table VII. — Primary myopathies associated with joint contractures

Disorder	Location/type of contractures	Reference
Muscular dystrophies		
– Duchenne muscular dystrophy	Hips, knees, ankles, hyperextension	(10,22,38,58,76)
– FSH	Location not mentioned	(57)
– X-EDMD (emerin)	Elbow, knee, Achilles tendon, cervical spine	(41)
– AD-EDMD (lamin A/C)	Elbow, knee, Achilles tendon, cervical spine	(6,41,71,98)
– EDMD non-specific (Hauptmann-Thannhauser MD)	Elbow, knee, Achilles tendons, cervical spine	(71,106)
– Congenital laminopathy	Proximal contractures, rigid spine	(106)
– LGMD 2A (calpain)*	Ankle	(42,45)
– LGMD 2B (dysferlin)*	Ankle	(90)
– Bethlem myopathy	Fingers, wrists, ankles, spine	(67,79,112)
– Ullrich CMD	Proximal > distal joints, tight Achilles tendon	(67,112,78)
– Fukuyama CMD	Hip, multiple	(121)
– Merosin-positive CMD	Hips	(13)
Congenital myopathies		
– Central core disease (RYR1)	Contractures, bone deformities	(360)
– Escobar syndrome (nemaline MP, TPM2)	Multiple pterygia	(23,84)
– Congenital fiber type dysproportion	Location not mentioned	(17,34)
– Multi-minicore MP	Cervical spine	(89)
– Multicore disease	Location not mentioned	(104)
– CMP with type 1 fiber predominance	Location not mentioned	(88)
– CMP with tubular aggregates	Achilles tendon	(28)
– CMP with ring fibers/vacuoles	Location not mentioned	(29)
– Arthrogryposis multiplex congenita (b-MHC)	Multiple joint contractures	(19)
– Arthrogryposis multiplex congenita (SYNE-1)	Multiple joint contractures	(1)
– Distal arthrogryposis type 1 (TPM2, TNNT3)	LL contractures, clubfoot, vertical talus	(52)
– Distal arthrogryposis type 2A (MYH7, TNNT3, TMP2)	LL contractures	(43)
– Distal arthrogryposis type 2B (TNNI2)	Limbs	(52,139)
– Distal arthrogryposis type 5	Location not mentioned	(3,150)
– Myosin MP (MYH2)	Congenital joint contractures	(96,140)
– Myosin MP (MYH3)	Distal arthrogryposis	(96)
– Myosin MP (MYH8)	Distal arthrogryposis	(96)
– Lipid raft disease	Location not mentioned	(53)
Distal myopathies		
– Distal MP with normal dysferlin	Achilles tendon	(87)
Myotonic syndromes		
– Congenital MD1	Hip abduction, knee	(10)
Metabolic myopathies		
– Glycogenosis V (McArdle)*	Location not mentioned	(74)
– Glycogenosis IV (Andersen)*	Ankle	(109)
Miscellaneous		
– Myopathy with muscle spindle excess (congenital)	Location not mentioned	(133)
– Beals-Hecht syndrome*	Multiple locations	(30)
– Rippling muscle disease (CAV3)	Achilles tendon	(75)
– Native American myopathy	Location not mentioned	(132)

LL : lower limb, * : variable childhood or adult onset.

pilot study on nine DMD patients the development of Achilles tendon contractures could be prevented by serial casting (*76*), which is applicable as long as the contractures are not fixed. As soon as contractures become fixed they can be solved only by surgical procedures for contracture release, such as arthroplasty, capsulotomy, aponeurectomy, or lower limb tenotomy (*131*). For severe knee contractures rotational osteotomy of the distal femur may be helpful (*47*). Recent advances also allow application of percutaneous correction of flexed hips, knees or equinus ankles (*47*). Hip contractures in DMD may be successfully treated by the modified Glorion-Rideau release (*39*).

Hyperlaxity and joint dislocation

Hyperlaxity is an increasingly recognised orthopaedic problem in patients with myopathy. Hyperlaxity is characterized by loose joint capsulae, ligaments and extreme non-physiological extension or flexion of joints. Hyperlaxity often results in dislocation or subluxation of a joint. Primary myopathies, in which hyperlaxity has been described include DMD, LGMD2E, CMDs, CMPs including arthrogryposis or rarely mitochondrial myopathy (Table VIII). Joints most frequently affected are the wrist and the hips. More rarely the knee is affected, such as in arthrogryposis, resulting in subluxation of the knee or subluxation or dislocation of the patella (patella alta) (*7*). Hyperlaxity of distal joints occurs particularly in CMD, such as Ullrich myopathy (*8*). Treatment of hyperlaxity is generally surgical and aims at correction of the dysplasia by various stabilizing methods (*47*). Procedures to treat or prevent subluxation or dislocation, however, are not indicated in asymptomatic patients and may be unsuccessful in maintaining a reduced hip (*47*). In case of acetabular deficiency and complex hip dysplasia triple pelvic osteotomy is used to optimize coverage and provides complete control over acetabular fragment placement (*156*). In case of superior or lateral dislocation of the patella quadricepsplasty and relocation of the patella may improve knee flexion (*7*).

Hand and foot deformities

Hand deformities

Hand deformities are an infrequent phenotypic feature of primary myopathies and concern the position of the fingers and the hand. Distal and proximal finger joints, metacarophalangeal joints

Table VIII. — Myopathies associated with joint hypermobility, hyperlaxity, subluxation, or dislocation (147)

Disorder	Reference
DMD (hip subluxation)	(12)
LGMD 2E (β-sarcoglycan)	(147)
Bethlem myopathy	(67,79,147)
Ullrich congenital muscular dystrophy	(32,67,147,112,78)
Merosin-positive CMD (luxation, dislocation)	(13)
CMD with joint hyperlaxity	(147)
CMP with type 1 predominance (hip dislocation)	(88)
Central core disease (hip dislocation)	(101,107,147,156)
Multi-minicore disease	(147)
Arthrogryposis multiplex congenita (luxation)	(7,108,154)
Spinal limitation (luxation)	(41)
CMP	(15)
Mitochondrial myopathy (hip dislocation)*	(136)

* : boy.

and carpo-ulnar joints may be affected alone or in combination. Myopathies with hand deformities include Bethlem and Ullrich CMD, distal arthrogryposis 5 (*3,150*) and multi minicore disease (*92*).

Tip-toe (plantar flexion)

One of the most frequent foot deformities in myopathy patients is the tip-toe. A tip-toe is characterized by fixed or flexible, non-physiologic plantar flexion of the feet with retention of the ability to stand on the heels. Tip-toes are most frequently due to an Achilles tendon contracture. They may not only occur isolated but often in combination with other foot deformities, in particular pes equinovarus. Primary myopathies, in which tip-toes have been described, are the dystrophinopathies, laminopathies, including EDMD and LGMD1B, caveolinopathies, dysferlinopathies, and distal arthrogryposis (Table IX). About one quarter of the DMD patients develop achillo-tendon contracture during the disease course (*50*). Treatment of tip-toes as a chronic Achilles tendon disorder is usually surgical. Only occasionally conservative measures, like night splints in DMD patients, have been tried (*50*). Various surgical procedures are utilized for tendo-Achilles lengthening, such as flexor hallucis longus transfer, posterior tibial-tendon transfer, proximal gastrocnemius recession, tenotomy of the Achilles tendon, or endoscopic recession (*14*).

Pes equinovarus (clubfoot, talipes)

The most prevalent of the foot deformities in primary myopathies is pes equinovarus, also known as clubfoot or talipes. Talipes is defined by the presence of three characteristic features : tip-toe, varus position (supination) of the subtalar foot, and inward rotation of the subtalar foot. Because of the talar rotation in the horizontal plane subluxation or luxation of the navicular occurs. Secondary contractures of the plantar fascia, the calcaneofibular band, the tendons of the long flexor of the hallux and the long digitorum flexor, and of the calcaneonavicular band develop. Foot deformities are frequently combined with other orthopaedic abnormalities, such as contractures, or pelvic abnormalities. Primary myopathies associated with talipes are listed in Table X. Treatment of choice includes conservative measures combined with surgical methods. Only if conservative measures are ineffective and the deformity is progressive, are surgical measures indicated. Among the surgical approaches the Ponseti regimen is the most widely applied. The Ponseti method and related methods aim at redressing and subtalar derotation. This is achieved by changing plaster casts, tenotomy of the Achilles tendon, canes (Denis Brown Bar), and anterior tibial muscle transfer. Another surgical technique is the complete subtalar release according to McKay-Simons (*148*). A surgical option in untreated congenital talipes from AMC is double zigzag incision (*56*). In case of complex relapses soft tissue release by posterior-medial and lateral ankle release, Achilles tenotomy, talonavicular capsulotomy, or reposition with K-wire fixation, are applied (*62*). Despite adequate treatment clubfeet have a tendency to relapse. Other foot deformities observed in patients with myopathy include the pes planovalgus (flatfoot) (*69*), the pes cavovarus (*63*), and the rockerbottom foot deformity (*23*).

Table IX. — Myopathies with toe-walking

Disorder	Reference
DMD	(50,131)
BMD	(18)
AD-EDMD (lamin A/C)	(6)
EDMD	(138)
Laminopathy	(110)
Rippling muscle disease (CAV-3)	(75)
Distal myopathy dysferlin-positive*	(87)
Distal arthrogryposis 5	(150)

* : 7 yo boy.

Differential causes of orthopaedic abnormalities

Orthopaedic abnormalities described above are not only a consequence of myopathies but may also be due to other causes, such as CNS disorders, neuropathies or neuronopathies, secondary muscle disorders, or primary orthopaedic disorders. Secondary orthopaedic disease in CNS disorders particularly occurs if there is weakness or spasticity.

Table X. — Myopathies with foot deformity (clubfeet, talipes (pes equinovarus), or pes planovalgus (flatfoot)

Disorder	Reference
Muscular dystrophy	
– DMD (talipes)	(117)
– EDMD	(86)
– Laminopathy (talipes)	(106)
– Ullrich myopathy	(79)
– Bethlem myopathy	(79)
– CMD (LMNA)	(106)
– Merosin-deficient CMD	(9)
Congenital myopathies	
– Central core disease (clubfeet)	(69,156)
– Multicore disease (clubfeet)	(104,149)
– Congenital fiber-type disproportion	(149)
– Centronuclear myopathy	(149)
– Minimal change myopathy	(143)
– Actin-myopathy (ACTA1)	(40)
– Distal arthrogryposis 5 (pes cavus)	(150)
– Distal arthrogryposis	(51)
– AMC* (clubfeet)	(43,8587)
Myotonic myopathies	
– Congenital MD1 (clubfoot, talipes)	(10,155)
Metabolic myopathy	
– MERRF syndrome	(145)

* : Arthrogryposis multiplex congenita.

CNS disorders causing weakness and wasting and secondary orthopaedic problems include stroke, infection, immunologic disease such as multiple sclerosis, trauma, surgery, tumour, or genetic disease like hereditary spastic paraplegia. Neuropathies, which secondarily develop orthopaedic disease include spinal muscular atrophy or amyotrophic lateral sclerosis. Primary orthopaedic abnormalities that need to be included as differentials include osteoarthritis, synovial inflammation and swelling, or damage to joint cartilages or to intervertebral discs. Additionally, immobilization from osteoporosis or other causes may facilitate the development of secondary orthopaedic disease.

CONCLUSIONS

Orthopaedic abnormalities of the spine, thorax, shoulder girdle, pelvis, and extremities may be a dominant phenotypic feature of various primary myopathies (table XI). These orthopaedic abnormalities most frequently occur in muscular dystrophies, CMPs, myotonic myopathies, and various unclassified myopathies. Only occasionally, orthopaedic abnormalities develop in myofibrillar myopathies, distal myopathies, or metabolic myopathies. Treatment at the right time has a strong impact on the prognosis and outcome of affected patients, since stabilization of the spine and correction of spinal or thoracic deformities may markedly

Table XI. — Myopathies with secondary orthopaedic abnormalities

Myopathy	DHS	SCO	HHR	SCA	TXA	CTC	FOD
Muscular dystrophies							
DMD	-	Y	Y	Y	Y	Y	Y
BMD	-	Y	-	-	-	-	Y
FSH*	Y	Y	Y	Y	Y	Y	-
EDMD	-	-	Y	Y	-	Y	Y
LGMD1B (laminin)	Y	Y	-	Y	-	Y	Y
LGMD 2A*	-	-	Y	Y	-	Y	-
LGMD2B*	-	-	-	-	-	Y	-
LGMD2C (gamma-sarcoglycan)	-	-	Y	Y	-	-	-
LGMD2D (alpha-sarcoglycan)	-	-	-	Y	-	-	-
LGMD2G (telethonin)	-	-	Y	Y	-	-	-
Rippling muscle disease	-	-	-	-	-	Y	Y
Ullrich myopathy	-	Y	Y	-	-	Y	Y
Bethlem myopathy	-	Y	-	-	-	Y	Y
CMD (lamin A/C)	Y	-	-	-	-	Y	Y
Fukuyama CMD	-	-	-	-	-	Y	-
Merosin-deficient CMD	-	-	-	-	-	-	Y
Merosin-positive CMD	-	-	-	-	-	Y	-
Rigid spine syndrome	-	Y	Y	-	Y	-	-
Congenital myopathies							
Central core disease	-	Y	Y	-	-	Y	Y
Nemaline myopathy	Y	-	-	-	Y	-	-
Escobar syndrome	-	Y	-	-	-	Y	-
Multicore disease	-	Y	Y	-	Y	Y	Y
Multi-minicore disease	-	Y	Y	-	Y	y	-
CMP with type I-predominance	-	Y	Y	-	-	-	-
Congenital fiber-type disproportion	-	Y	-	-	-	Y	Y
Centronuclear myopathy	-	-	--	-	Y	-	Y
Minimal change myopathy	-	-	-	-	Y	-	Y
Actin-myopathy (ACTA1)	-	-	-	Y	Y	-	Y
Arthrogryposis multiplex congenita	-	Y	-	-	Y	Y	Y
Distal arthrogryposis	-	-	-	-	Y	Y	Y
Myosin myopathy	-	-	-	-	-	Y	-
Reducing body myopathy	-	Y	Y	-	-	-	-
Cap-myopathy	-	-	-	Y	-	-	-
Myofibrillar myopathies*	-	Y	-	-	-	Y	-
Distal myopathies							
Distal myopathy dysferlin-positive	-	Y	-	-	-	-	Y
Myotonic myopathies							
MD1#	Y	Y	-	-	-	-	-
MD2#	Y	-	-	-	-	-	-
Congenital MD1	-	Y	Y	-	-	Y	Y
Metabolic myopathy							
Glycogenosis II (acid maltase deficiency)	Y	-	Y	Y	-	-	-
Glycogenosis IV*	-	-	-	-	-	Y	-
Glycogenosis V*	-	-	-	-	-	Y	-
Lipid raft disease	-	-	-	-	Y	Y	-
Mitochondrial myopathies*	Y	-	Y	-	-	-	Y
Miscellousnous							
Myopathy with muscle spindle excess	-	-	-	-	-	Y	-
Beals syndrome*	-	-	-	-	-	Y	-
Native American myopathy	-	Y	Y	-	-	Y	-
Scapuloperoneal syndrome#	-	-	Y	-	-	-	-
Valosin-containing myopathy*	-	-	-	Y	-	-	-

DHS : dropped head syndrome, SCO : scoliosis, HHR : hyperlordosis, hyperkyphosis, rigid spine, SCA : scapula alata, TXA : thoracic abnormalities, CTC : contractures, FOD : foot deformities, y : yes, * : variable childhood or adult onset, # : adult onset.

improve pulmonary and cardiac functions and may be the prerequisite for effective non-invasive positive pressure ventilation. Orthopaedic surgery may additionally provide improved mobility, stability and function of arms, legs, hands, and feet and may be the basis for successful physiotherapy. If invasive measures are not indicated or are contraindicated, conservative measures including physiotherapy, canes, orthoses, splints, or casts may be beneficial. Since still little is known about the frequency, familial distribution, phenotypic heterogeneity of orthopaedic abnormalities in primary myopathies collection of more data, particularly in genetically confirmed myopathies is warranted. To improve the management of myopathy patients with orthopaedic problems, a multidisciplinary diagnostic and therapeutic approach with more intense cooperation between neurologists, orthopaedists, anesthesiologists, physiotherapists, bandagists, truss makers, and kinesiologists is required.

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