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Rehabilitation in Muscular Dystrophies: Changing Approach

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1. Introduction

Life expectancy is increasing in muscular dystrophies: as an example due to technical medical interventions like spine surgery and home ventilation, boys with Duchenne muscular dystrophy become men. Increasing evidence concerning retarding drugs becomes available for muscular dystrophies. Cardiac symptoms can effectively be treated with drugs and also cardioprotective drugs are tested. New retarding treatments, like exon skipping, stem cell treatment or vector-gene transfer, are in the phase of animal studies or already in randomized clinical trials. Together these treatments are changing the course of the muscular dystrophies.

In line with these developments rehabilitation management is also changing. In the past the treatment was focused on maintaining walking abilities as long as possible with physiotherapy, stretching, and with or without braces. However, due to the slower progression of the disorder and the technical possibilities of home ventilation the focus is changing to arm- and hand-function. There are good technical solutions for the loss of ambulation as there are many type of electrical wheelchairs. All kind of technical and electronic supports are available to operate telephone, television, radio etc. However, in all kind of daily activities a certain ability of the arm and hand is necessary. Training and support of these functions in muscular dystrophies come into prominence.

New symptoms or symptoms not noticed in the past are becoming more apparent. This can be due to the increasing age that they are now identified or augment during the longer course of the disease, such as feeding and swallowing problems, gastrointestinal and urogenital problems.

Already known symptoms like osteoporosis in muscular dystrophies also increase if corticosteroids are used. For example vertebral fractures are more seen in boys with Duchenne muscular dystrophy. The question is whether we can retard osteoporosis by means of supported physical activities with weight load on the bones. In the light of increasing life expectancy and the possibilities to use sophisticated orthoses or exoskeletons, it is important to maintain the physical capacities with balanced training.

Also due to increasing life expectancy an unforeseen population emerges who want to participate in social life in broad sense: education, jobs, friends, relationships, marriage,

children. This desire is in slight contrast to adults who received the diagnosis muscular dystrophy in adulthood, they are attempting to maintain their participation in social life. Possibilities to live independent with supportive devices need to be established, robotics as support in daily activities will develop, and with the possibilities of the world wide web and computer technology education and jobs become available.

In this chapter we will not describe the established rehabilitation managements, but will describe the new focuses on training, the new or less noticed symptoms, and the development of new devices.

2. Training

2.1 Physical training for children with a muscular dystrophy

2.1.1 Disuse

Muscular dystrophies in children comprise a heterogeneous group of myopathies that appear during childhood. The most common muscular dystrophy in children is Duchenne Muscular Dystrophy (DMD) affecting 1/4200 life-born boys. Other muscular dsytrophies are Myotonic Dystrophy (MD), Becker Muscular Dystrophy (BMD), Limb Girdle Muscular Dystrophy (LGMD) and FacioScapuloHumeral muscular Dystrophy (FSHD). All muscular dystrophies are characterized by progressive loss of muscle function and only symptomatic treatments (such as corticosteroids and assisted mechanical ventilation) are currently available. An important aim in the management of muscular dystrophies is to delay the loss of functional abilities and to maintain independency. Physical training could retard the loss of physical abilities as a result of disuse.

Children with a muscular dystrophy are less physically active compared to age-matched healthy controls in their daily life (McDonald et al 2005). The more sedentary lifestyle can be explained primarily by the disease, but also by disuse(McDonald 2002, Bar-or and Rowland 2004). Disuse can be defined a discrepancy between children's capacity and performance, and gradually causes a secondary reduction of physical activity. Indeed, the increasing amount of energy a certain activity costs, and the fear of falls with the need for help to stand up, make children move less. An early decline of the physical activity level enhances the loss of functional abilities. This appears from the loss of arm functions that occurs fast after the onset of wheelchair-dependency(McDonald et al. 1995). An electric wheelchair limits arm functions (like lifting and reaching), since a top blade and a central operating joystick force children to function within the outlines of the wheelchair. Another example is the high number of children with DMD (20-40%) that loses the ability to walk as a result of a fracture of the lower extremity(Vestergaard et al , 2001). From this perspective, the saying "use it or lose it" is certainly applicable to children with a muscular dystrophy and encourages physical training.

2.1.2 Evidence for training

The number of studies that investigated the effects of physical training in children with a muscular dystrophy is limited (Voet et al. 2010). None of them were performed in a randomized controlled setting and most studies focused on DMD. Furthermore, clinical trials in children only investigated the effect of resistance exercises. This is remarkable, as

recent studies in adults with BMD and the mdx mice (a mouse model for DMD) encourage aerobic exercises.

Previous clinical trials among boys with DMD showed that (sub)maximal resistance exercises have limited positive effects on muscle strength and time functional tests (such as the time it takes to walk 10m) but, importantly, are not harmful. In a study by Vignos et al. (1966), the effects of a one-year maximum resistance exercise program (i.e. the maximum load that could be lifted through ten repetitions) were examined(Vignos, Jr. and Watkins, 1966). Fourteen ambulatory children with DMD exercised their legs, arms and abdominal muscles and were compared with a control group of children with DMD who did not exercise. Results from this study showed that muscle strength decreased in control group, while strength was maintained during the training period in the exercise group. However, children were not randomly allocated to the exercise or non-exercise (natural cohort) group. Another study by De Lateur et al. (1979) showed that a six-month submaximal isokinetic exercise program could be of limited value in increasing strength in DMD (de Lateur and Giaconi, 1979). In this study, four ambulatory children with DMD performed quadriceps exercises with one leg (4 to 5 days per week), while the other leg was not trained at all. Finally, Scott et al. (1981) investigated the effects of six months manually applied resistance exercises and "free exercises" in eighteen boys with DMD(Scott et al., 1981). At six months, no statistically differences were found between the two groups at the level of muscle strength, locomotor abilities and functional abilities. No evidence for training-induced physical deterioration was found.

With respect to aerobic exercises, recent studies in mdx mice (an animal model for DMD) showed that voluntary wheel running had positive effects on muscle strength and fatigue resistance and non-weight bearing exercises (such as swimming) had no detrimental effects 1998; Hayes and Williams 1996). Dynamic exercises (bicycle training) improved endurance and muscles strength in adult BMD patients as well(Sveen et al. 2008) . In this study of Sveen et al. (2008), eleven ambulatory BMD patients, and seven healthy age-matched controls, participated in a 12-weeks cycling training. Participants cycled thirty minutes at 65% of their maximal oxygen uptake (VO_{2max}). At 12 weeks, workload and VO_{2max} were improved without an increase in CK level. Although this study was conducted among adults, the results of this study might be applicable for children with a muscular dystrophy as well.

Results of recently published clinical trial protocols, such as the protocol of the randomized controlled trials No Use is Disuse (NUD)(Jansen et al. 2010), will increase insight into what type of physical training (type, intensity, frequency, duration) should be recommended to children with a muscular dystrophy. The NUD study investigates whether an assisted bicycle training is beneficial and does not cause any harm for boys with DMD. Motor-assistance allowed cycling with the legs and arms (arm cranking) even when muscle strength was insufficient to achieve fully active movements. In another part of the study, the effect of an arm training with arm support is investigated. Both ambulatory and wheelchair-dependent children participate in this study. Preliminary data show that assisted training is effective in boys with DMD in maintaining functional capacities.

2.1.3 Training mechanisms

The mechanism by which training could oppose the physical deterioration in children with a muscular dystrophy is still unclear. Muscle fibers in muscular dystrophy patients are

abnormally vulnerable to contraction-induced injury due to the absence, or lack, of mechanical reinforcement of the sarcolemmal membrane(Petrof 1998). Eccentric exercises should therefore be avoided(Lim et al 2004). Conversely, a recently published review by Markert et al. (2011) described that enhancing myofiber repair, decreasing muscle fibrosis and the production of antioxidants against oxidative damage are potential explainable factors for exercise-induced improvements(Markert et al. 2011). Work-induced damage could enhance muscle regeneration and repair(Okano et al. 2005), and low-stress exercise may produce beneficial effects on myofiber contractility and energetic efficiency(Petrof 1998).

2.1.4 International training guidelines

International training guidelines for children with a muscular dystrophy are preferably disease-specific and should be adapted to the individual child(Edouard et al. 2007). Based on the currently available evidence, guidelines for ambulatory boys with DMD recommend voluntary active exercises (such as swimming) and to avoid eccentric exercises(Eagle 2002). For wheelchair-dependent children, passive or actively-assisted mobilizing exercises to maintain comfort and symmetry are advised(Eagle 2002). A cardiomyopathy could be a contraindication to participate in any physical training programs(Edouard et al. 2007). It is suggested that the training intensity could be based on children's perceived exertion instead of maximum heart rate when performance is predominantly limited by the peripheral capacity instead of the oxygen transport(Jansen et al. 2010).

To conclude, physical training could delay the physical deterioration as a result of disuse in children with a muscular dystrophy. Disuse enhances the loss of functional abilities, whereas physical training could be beneficial. Currently available evidence is limited to uncontrolled clinical trials and further research is required to develop specific training prescriptions. At this moment, international guidelines recommend voluntary (dynamic) exercises to maintain comfort and symmetry, and to avoid eccentric exercises.

2.2 Training and fatigue in adult muscular dystrophies

2.2.1 Physical exercise

In a study by McDonald the three problems most frequently cited as "very significant" by patients with slowly progressive neuromuscular disease were muscle weakness (57%), difficulty exercising (43%) and fatigue (40%) {McDonald 2002}. Two main types of fatigue can be distinguished. Physiological fatigue, or muscle fatigue, has been defined as a reduction in maximal voluntary muscle force (MVC) during exercise. Experienced fatigue, on the other hand, is the subjective feeling of fatigue. Muscle fatigue is not necessarily accompanied by experienced fatigue, or vice versa. Distinguishing experienced fatigue from muscle weakness, the key feature in muscular dystrophy, may be difficult. In a study by Kalkman 61% of patients with facioscapulohumeral dystrophy (FSHD) (n = 139) and 74% of patients with myotonic dystrophy (MD) (n = 322) were "severely fatigued" {Kalkman et al, 2005 }. Patients with MD had higher scores for experienced fatigue, reported greater problems with concentration and had more difficulties with initiative and planning than patients with FSHD. In FSHD patients and MD patients, social functioning was related to fatigue severity. Apparently, fatigue is not only a frequent, but also a relevant problem in

muscular dystrophy. In a subsequent longitudinal study Kalkman built a model of perpetuating factors, which contribute to the continuation of experienced fatigue {Kalkman et al, 2007 }. In FSHD, the level of physical (in)activity has a central place in the model. Due to fatigue, patients often alter their lifestyles and reduce their activities. Low physical activity levels may lead to even greater weakness and atrophy of skeletal muscles, which causes a vicious circle of disuse and weakness. Physical inactivity in turn can lead to chronic cardiovascular and muscle deconditioning and increased cardiovascular health risks {McDonald, 2002 }. In addition, pain complaints influence levels of experienced fatigue both directly and indirectly by decreasing physical activity (see figure below). In MD, physical activity and pain did not significantly contribute to experienced fatigue. Yet, sleep disturbances lead to higher levels of experienced fatigue in both FSHD and MD patients. The observed patterns of perpetuating factors can be used as a basis to develop evidence-based interventions to reduce fatigue. Specific attention should be paid to sleep disturbances in both patient groups. Specifically in FSHD, treatment of fatigue should also be directed at increasing physical activity and reducing pain complaints. Irrespective of its cause, physical inactivity should be discouraged in muscular dystrophy patients because of an increasing risk of cardiovascular disease and muscle deconditioning.

In the past, many patients with muscular dystrophy were advised not to exercise because of the belief that too much exercise might lead to overuse weakness {Johnson, 1971;Johnson, 1971;Carter, 1995;Fowler, 1984;Petrof, 1998}. Yet, in their Cochrane review on muscle strength training and aerobic exercise training for patients with muscle diseases, Voet concluded that moderate-intensity strength training in MD and FSHD appeared not to be harmful, although there was insufficient evidence to establish its benefit {Voet et al , 2010}. This conclusion was based on merely two randomised clinical trials {Lindeman et al , 1995;van der Kooi et al, 2004}. For this reason, Cup reviewed not only randomised clinical trials, but also controlled clinical trials and other designs of sufficient quality {Cup et al, 2007}. All types of exercise therapy and other physical therapy modalities were included for patients with muscular dystrophy, among which were patients with FSHD, LGMD, MD and DMD. Cup *et al.* also concluded that exercise training is not harmful in muscular dystrophies. However, based on the reviewed studies, there was insufficient evidence for the effectiveness of muscle strengthening exercises, although there were some indications that aerobic exercises may have a positive effect on body functions, as well as on activities and participation. Because of the weakness of the muscle membrane there is concern about the potentially damaging effects of eccentric and high-intensity muscle contractions during strength training. In animal models of muscular dystrophy, there is evidence that eccentric contractions, known to stress muscle fibers, cause greater cell injury to these dystrophic muscle fibers. Although transferring results from animal studies to humans must be done with caution, eccentric training studies in muscular dystrophy patients are so far being avoided.

To conclude, although the current scientific evidence is scarce, aerobic exercise training appears not to be harmful in muscular dystrophies and could have a positive effect on functioning, activities and participation, but the number of high-quality studies is low. When prescribing exercise training, the recommendations from the ACSM Position Stand can be used as requirements to achieve an effective, safe and individualised exercise prescription {1998}.

2.2.2 Alternative training

Muscular dystrophies have a large impact on psychosocial functioning as patients must continuously adapt to their progressive illness. Illness cognitions and coping styles influence the level of physical activity and, consequently, experienced fatigue and health status. Hence, changing illness cognitions and coping style may lead to a better quality of life. A cognitive behaviour approach has been proven successful in the chronic fatigue syndrome {Prins et al, 2001;Chambers et al, 2006} and for post-cancer fatigue {Gielissen et al, 2007;Gielissen et al, 2006} and may be effective in patients with muscular dystrophy as well.

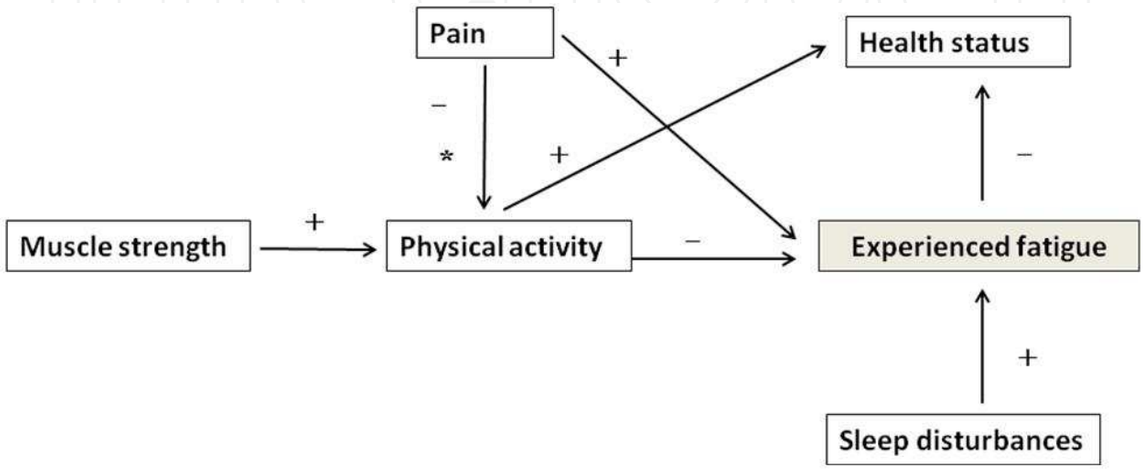

Fig. 1. Cognitive behaviour therapy in FSHD should, for instance, be focused on the known perpetuating factors of experienced fatigue as described by Kalkman i.e. sleep disturbances, pain complaints and physical inactivity (Della et al. 49-53;Kalkman et al. 571-79). Therapy should be adapted to the life of each individual, resulting in an individualised treatment approach. Altogether, cognitive behaviour therapy seems a rational, promising treatment for fatigue in muscular dystrophies.

3. Feeding problems and dysphagia

3.1 Swallowing

Swallowing is a complex sensorimotor process that depends on information from multiple levels of the central and peripheral nervous system. Descending excitatory and inhibitory signals from the cortex and subcortex and ascending signals from the oropharyngeal area trigger the central pattern generator (CPG) in the bulbar reticular formation (Jean 2001, Leopold 2010). This network of premotor neurons and interneurons drives the motor neurons of swallowing in cranial nerves (CN) V, VII, IX, X and XII. Muscles of lips, submental muscle group, tongue, palate, larynx, pharynx and esophagus, innervated by these CNs, are excited and inhibited sequentially, when a person forms a bolus and swallows (Ertekin 2003). The oral phase and the initiation of the pharyngeal phase are under voluntary neural control, whereas the completion of the pharyngeal phase and the esophageal phase are under involuntary neural control (Miller 2008). In dysphagia, problems may occur in the oral, pharyngeal, esophageal phase, or in more than one phase (Arvedson 2008). In children with neurologic etiologies from different origins, dysphagia is often reported with variable signs and

symptoms. The dysphagia of children with CP is characterized by oral motor problems and frequent aspiration of thin liquid with no observable response (silent aspiration) (Rogers 1994). In neuromuscular disorders (NMD), weakness of the muscles is due to damage along the course of the peripheral nerve (lower motor neuron) or the muscle itself (Dubowitz 2000). Feeding and swallowing problems are not uncommon in muscular dystrophies, which can lead to dehydration, malnutrition or aspiration pneumonia.In patients with chronic muscle disease a prevalence of 35% of feeding problems and dysphagia was reported (Kumin 1994).

3.2 Problems in the oral phase

In NMD chewing problems and the need to multiple swallows to clear the oral cavity (piecemeal deglutition) are the main problems in the oral phase of swallowing. Reduced bite force and weakness of the masticatory muscles (Morel-Verdebout 2007) causes chewing problems and the inability to eat solid or firm food. The reduced range of mandibular motion, reported in SMA type II (Van Bruggen 2011) and DMD (Botteron 2009) may reduce the quality of chewing and results in a hampered food comminution, inadequate food bolus formation, and oral transport. Moreover, the limited mandibular range of motion has an impact on oral hygiene and dental care. Facial muscle weakness can lead to craniofacial and dental malocclusion (Kumin 1994; Pane 2006) which aggravates the chewing problems.

Reduced strength of the tongue muscles causes piecemeal deglutition, especially with thick liquid and solid food (van den Engel-Hoek 2009). This results in prolonged oral transit time and oral residue after swallowing.

3.3 Problems in the pharyngeal phase

The initiation of the pharyngeal phase is typically normal in patients with NMD, but problems with solid food and residue after swallow are reported in patients with SMA type II (van den Engel-Hoek 2009), DMD (Aloysius 2008;Shinonaga 2008), in MD (Leonard 2001) and congenital myopathies (Mehta 2006;de Swart 2002). In all patient groups the problems occurred more in advanced stages. Dysphagia and feeding problems were also reported in nemaline myopathy, but more in the neonatal period and infancy than in adults (Bagnall 2006). Residue after swallow is caused a combination of reduced strength of the tongue and submental muscle group, and reduced opening of the upper esophageal sphincter. In patients with oculopharyngeal muscular dystrophy (OPMD) the dysphagia was aggravated by the retroflexion of the head, caused by ptosis (de Swart 2002). In patients with SMA II a retracted position of the head, due to a lumbar lordosis and a diminished head balance, caused reduced movement of the submental muscle group resulting in more post swallow residue with thick liquid and solid food than with thin liquid. The post swallow residue places patients at risk for aspiration the airway reopens (Arvedson 1998)

3.4 Swallowing assessments

A videofluroscopic swallow study (VFSS) is generally considered as the gold standard method for diagnosing dysphagia in adults (Logemann 2000) and children (Hiorns 2006), especially to detect aspiration. However, a VFSS was not considered as additional benefit to a careful feeding history in patients with DMD (Aloysius 2008). Other assessments are available to describe oral structures and biomechanical oral functions needed for feeding

and swallowing to better understand the nature and clinical course of dysphagia in NMD. Surface EMG (sEMG) of the submental muscle group, tongue pressure, ultrasonographic and manometric assessments during swallowing can be used to understand the nature and clinical course of dysphagia in NMD.

Patients are not always mentioning swallowing difficulties (Leonard 2001;Stubgen 2008). They should also always be carefully interviewed for symptoms of swallowing difficulties that may require a swallowing assessment and a careful observation of oral motor abilities during mealtime (Messina 2008; Manzur 2008).

3.5 Recommendations

To improve their quality of life and nutritional state, NMD patients with swallowing difficulties benefit from dietetic and swallowing recommendations. This can be nutritional, related to consistencies, safe swallowing techniques or advices about postural management and feeding aids . The dysphagia in NMD show different patterns in flaccid bulbar paresis like NMD than in neuromuscular disorders. In contrast to the usual advice for thickening the food, in NMD more liquid food is advised or alternating thick with thin consistencies. Strategies are recommended to reduce problems in chewing and oral and pharyngeal post swallow residue. Careful chewing and bolus preparation to a liquid consistency, effortful swallowing and double swallows can reduce problems of reduced pharyngeal clearing. In case of post swallow residue it is also advised to clear the oral and pharyngeal cavity with water after mealtime. Positioning of the head in sitting position can also be important to prevent residue. In case of a retracted head position, like in SMA II (van den Engel 2009) a more flexed head position prevented laryngeal post swallow residue. Also in patients with OPMD a slightly flexed head improved swallowing (de Swart 2002).

4. Gastrointestinal problems

If swallowing and chewing gets more difficult, eating takes too much time or energy, enteral tube feeding is a possibility. The percutaneous gastrostomy is quite regular, although there are also complications described during (re)placement. In myotonic dystrophy impairment of gastrointestinal motility is known and seems to be gradual worsening(Bellini et al 2006), probably related to gastrointestinal symptoms like regurgitation, dyspepsia, abodominal pain, bloating, and changes in bowl habits. Although there is only a low correlation between the degree of skeletal muscle involvement and the presence and severity of gastrointestinal disturbances it is a positive correlation (Bellini et al 2006). Also in boys with Duchenne and Becker muscular dystrophy abnormalities in gastric motility are seen and the possibility of progressive failure in neuromuscular function is put forward (Borelli et al 2005). Promotility agents are advised as are good diet, stool softeners, and hydration (Bellini et al 2006, Wagner et al 2007). There is little evidence on the benefits of exercise and chronic constipation (Leung et al 2011), but practical experience often reported is that doing exercise or standing in a standing table or frame does seem to help. This would be in line with the former recommendations for exercise.

5. Urogenital problems in muscular dystrophies

Lower urinary tract symptoms are described in boys with Duchenne muscular dystrophy and seem not to be rare (van Wijk et al 2009). Nearly 85 % of the boys/men with Duchenne

muscular dystrophy reported lower urinary tract symptoms, and 51 % had more than 3 problems. The main problems were post micturation dribble, straining, (urge) incontinence, and feeling of incomplete emptying. There is a very low correlation between age and functional abilities and lower urinary tract problems; these problems seem not to worsen with disease progression. In 42 % the boys/men mentioned that the complaints influenced their social life, and 25 % reported a decrease in quality of life. Also in myotonic dystrophy urinary tract symptoms are reported but only anecdotal. This is probably an underreported symptom and should have attention in the management.

It is recommended to ask for lower urinary tract symptoms, use a micturation questionnaire if there are concerns, and register for several days the pattern. Up till now no pathophysiological studies are available, the possibility of a bladder-sphincter dys-synergy is suggested, and the symptoms are treated symptomatically by medication.

6. Technical possibilities

New very sophisticated devices are being developed like exoskeletons, robot arms and motion controlled orthoses (see for example websites of www.Flextension.nl and www.FocalMeditech.nl). Not only can these devices support a natural function, but they can also simulate a natural movement. It is possible that with an exoskeleton a person with severe paresis can still move in a rather natural way. In an earlier stage these devices can also be used for assisted training and it is theoretically possible that by regular use one can prevent contractures. Robotica can potentially reduce the extent of personal help needed, thus making an adjusted independent life possible and making participation in social life possible.

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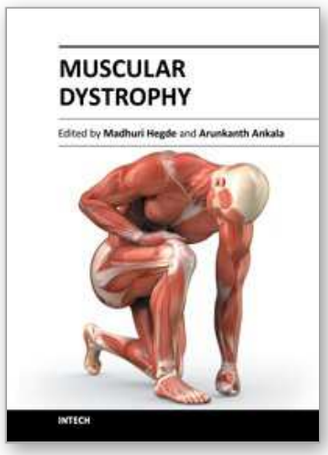

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With more than 30 different types and subtypes known and many more yet to be classified and characterized, muscular dystrophy is a highly heterogeneous group of inherited neuromuscular disorders. This book provides a comprehensive overview of the various types of muscular dystrophies, genes associated with each subtype, disease diagnosis, management as well as available treatment options. Though each different type and subtype of muscular dystrophy is associated with a different causative gene, the majority of them have overlapping clinical presentations, making molecular diagnosis inevitable for both disease diagnosis as well as patient management. This book discusses the currently available diagnostic approaches that have revolutionized clinical research. Pathophysiology of the different muscular dystrophies, multifaceted functions of the involved genes as well as efforts towards diagnosis and effective patient management, are also discussed. Adding value to the book are the included reports on ongoing studies that show a promise for future therapeutic strategies.

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