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(CZ) Vývoj rehabilitace a léčby Duchennové svalové dystrofie: narativní přehled.

(EN) The evolution of rehabilitation and treatment in Duchenne muscular dystrophy: a narrative review.

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SÚHRN/ABSTRAKT

Východisko: I přes moderní technologie a léčbu je Duchennova svalová dystrofie onemocnění, které vede v útlém věku k fyzické degradaci organismu až k jeho smrti.
Súbor: Cílem práce je předložit výsledky o vývoji léčby a rehabilitace pacientů s Duchennovou svalovou dystrofií a definovat aktuální možnosti rehabilitace k udržení co nejdelšího stavu samostatnosti a soběstačnosti.
Metódy: Analýza dostupných dat a přehled zdrojů o léčbě a rehabilitaci Duchennovy svalové dystrofie se zvláštním zřetelem na fyzioterapii byla provedena ke dni 31. května 2025 z databází Národní lékařské knihovny, Univerzitní knihovny ČVUT, Medvik, Scopus, PubMed (Medline) a Web of Science. Vyhledávání bylo provedeno primárně na základě stanovených klíčových slov.
Výsledky: Rehabilitace u DMD musí být postavena na komplexním přístupu. Úspěšnost fyzioterapie závisí na časném začátku, individuálním přístupu, kontinuitě a úzké mezioborové spolupráci. Mimo pohybového aparátu musí být zaměřena péče i orofaciální a dechové funkce, kardiovaskulární systém a psychosociální stabilitu pacienta i jeho rodiny. K využívání prvků z klasických terapeutických konceptů, balneologie, hydrokineziologie se ukazuje vhodné zapojení telerehabilitace z důvodu zachování dostupnosti a kontinuity péče.
Závery: Historický přehled ukazuje na složitosti dřívější diagnostiky a omezené možnosti terapie. Přestože se stále jedná o nevyléčitelné onemocnění, současné klinické a rehabilitační přístupy umožňují významně ovlivnit průběh nemoci. Do budoucnosti se předpokládá, že tzv. genová terapie bude zásadním milníkem, který povede k změně ve zdraví osob DMD/BMD.

SUMMARY/ABSTRACT

Starting point: Despite modern technology and treatment, Duchenne muscular dystrophy is a disease that leads to the physical degradation of the body at an early age, even leading to death.
Group: The aim of this paper is to present the results of the development of treatment and rehabilitation of patients with Duchenne muscular dystrophy and to define the current rehabilitation options to maintain the longest possible state of independence and self-sufficiency.
Methods: Analysis of available data and review of resources on treatment and rehabilitation of Duchenne muscular dystrophy with special reference to physiotherapy was performed as of 31 May 2025 from the databases of the National Library of Medicine, the University Library of CTU, Medvik, Scopus, PubMed (Medline) and Web of Science. The search was performed primarily on the basis of established keywords.
Results: Rehabilitation in DMD must be based on a comprehensive approach. The success of physiotherapy depends on an early start, individual approach, continuity and close interdisciplinary cooperation. In addition to the musculoskeletal system, the orofacial and respiratory functions, the cardiovascular system and the psychosocial stability of the patient and his/her family must be addressed. In addition to the use of elements from classical therapeutic concepts, balneology, hydrokinesiology, it appears appropriate to include telerehabilitation in order to maintain accessibility and continuity of care.
Conclusions: The historical overview shows the complexity of the earlier diagnosis and the limited therapeutic options. Although it is still an incurable disease, current clinical and rehabilitation approaches allow to significantly influence the course of the disease. In the future, gene therapy is expected to be a major milestone that will lead to a change in the health of DMD/BMD individuals.

KĽÚČOVÉ SLOVÁ

Duchenne muscular dystrophy, physical therapy, rehabilitation, myopathy, gene therapy.

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1 INTRODUCTION

Despite advanced modern medicine, there are diseases that we cannot yet influence at all, or only partially. These are mainly a group of genetically burdened diseases, which also include myopathies and muscular dystrophies. Only in recent years has gene therapy brought about a significant change, giving affected individuals the possibility of a higher quality of life.

Myopathies and muscular dystrophies are a heterogeneous group of neuromuscular diseases. Nevertheless, there are some common features of these diseases, which may include muscle weakness, especially of the brachial plexus muscle groups, as well as a flaccid posture, which is characterised by an accentuated lumbar lordosis and hypotonic abdominal bulging. Individuals affected by any of these disorders typically exhibit delayed motor skill development.[1–3]

The so-called Gowers' sign (myopathic climb) is visible in the context of uprightness, where the person suffering from this disorder is forced to use the support of the upper limbs when standing upright. On neurological examination, there is no noticeable impairment of surface sensation and, with the exception of occasional muscle spasms, patients do not experience significant muscle pain or myogenic pain, but it is not one of the dominant symptoms of the disease. As for myotatic reflexes, this depends on the stage of the disease, but they are usually reduced to absent. The clinical picture and progression of the disease is determined by the specific subtype of the dystrophy.[2,4]

Duchenne muscular dystrophy (DMD) is one of the more progressive subtypes of muscular dystrophies. It is a so-called dystrophinopathy, in which the gene responsible for the production of the dystrophin protein is mutated. This subtype of the disease is characterised by the complete absence of the functional form of this protein. Dystrophin is responsible for maintaining the structural integrity and functionality of muscle fibres. It forms the so-called dystrophin-glycoprotein complex, which links the inner cytoskeleton of muscle cells to the extracellular matrix and thus provides crucial support during muscle contraction and relaxation. In patients with Duchenne muscular dystrophy, this protein is absent, resulting in muscle cell death due to dysregulation of calcium homeostasis and oxidative damage. Thus, muscles are unable to contract physiologically, and muscle fibre repair is impaired.[5,6]

DMD occurs worldwide with an incidence of 1:3,500 male newborns. Female sex is not accounted for due to the type X-linked inheritance, where females are overwhelmingly carriers of the disease.[2,7] In addition to DMD, there is a milder form of the disease called Becker muscular dystrophy (BMD). This type is characterized by the presence of the dystrophin protein, but only in a dysfunctional form. The first clinical symptoms may appear as early as three years of age or during the individual's adulthood. Patients with this subtype also live into their fourth or fifth decade of life due to the slower development.[1,7,8]

2 THE DATA FILE AND METHODS

An analysis of available data was conducted and a review of resources on the rehabilitation of Duchenne muscular dystrophy was compiled with particular reference to physiotherapy.

On 31 May 2025, a search of the databases of the National Library of Medicine, University Library of CTU, Medvik, Scopus, PubMed (Medline) and Web of Science was performed to identify results used in published research on Duchenne muscular myopathy. Using the same databases, a secondary search was performed to find published scientific articles describing the history of the disease, treatment, rehabilitation and physiotherapy.

2.1 SELECTION STRATEGIES, SEARCH AND CRITERIA

To find research publications, the author team created search strategies for each database using terms related to the development of Duchenne muscular dystrophy rehabilitation. A keyword search was used to primarily include the scientific article: Duchenne, physiotherapy, rehabilitation, histo-array, myopathy, gene therapy. All possible combinations of keywords using AND and OR were used in the advanced search of all the databases mentioned.

No publication time interval was set for the keyword search of articles.

Secondary criteria for articles older than five years were monitoring the quality of the journal and the citation rate of the article.

Articles that did not address the issue of Duchenne muscular dystrophy were excluded. Duplicate publications were removed when comparing the different databases. Screening was always performed by two researchers.

Methods deviated from the a priori protocol in the inclusion criteria for articles. Specifically, articles from the "grey literature" were excluded.

3 RESULTS AND DISCUSSION

Clinical signs of the disease usually first appear around the third to fifth year of life. At first, there are difficulties with the development of quadrupedal locomotion. This stage may be skipped during motor development. Subsequent difficulties also relate to the development of bipedal locomotion itself, which usually occurs slightly later than in the healthy population. The individual often walks on tiptoe and has pathological gait abnormalities characterised by the typical sideways sway. In the case of the lower limb, a pelvic drop instead of an elevation is visible. The individual with DMD has marked difficulty walking up stairs, climbing any elevated surface or developing the skill of jumping. The pathology is accentuated within the faster walking to running gait. A significant symptom is the so-called pseudohypertrophy of the calf muscles. This condition is caused by the accumulation of adipose tissue between the muscle fibres of the calf muscles, which causes their increased volume and gives the appearance of hypertrophy. In reality, it is not a true overgrowth of muscle mass, but its replacement by adipose tissue, which thus causes dysfunction of the muscle in question. Around the age of 10, a person with DMD loses the ability to walk independently. They develop muscle contractures and spinal deformities (especially scoliosis). Respiratory muscle dysfunction and reduced mobility lead to higher rates of respiratory disease, resulting in reduced lung vital capacity and overall impaired respiratory function. Other major complications include cardiomyopathy, which usually manifests itself after the eighteenth year of life or later, and is usually asymptomatic. Individuals with DMD also often show delayed speech development and may develop various learning or cognitive impairments later in life.[1,2,9]

Muscle biopsy of skeletal muscle reveals degenerative processes occurring at the level of the muscle cell. These fibres are often visible as clusters and immaturity of the central nuclei can be observed. Under physiological conditions, these central muscle fibres have a significant influence on muscle regeneration. Within the muscle fibre, myoblasts provide this regenerative capacity, especially in the early phase of the disease, ensuring homeostasis between ongoing necrotic and regenerative processes. Because of the establishment of this imbalance and the subsequent predominance of degenerative processes, muscle fibres are remodelled and become less resistant. They are particularly at risk in eccentric contraction, where the stretching of the muscle fibre increases the demands on the already fragile muscle membrane severalfold. This can lead to the formation of micro lesions, which in large quantities can cause loss of calcium ions and even cell death.[10,11]

3.1 MOST COMMON COMPLICATIONS OF DUCHENNE MUSCULAR DYSTROPHY

In the development of DMD, health care professionals and family members must take into account the possible development of complications. Due to the progressive decrease in muscle strength, the proximal and respiratory muscles gradually weaken, leading to more rapid fatigue. Difficulties with expectoration of mucus and its accumulation are associated with respiratory muscle impairment, and nighttime and later daytime hypoventilation with the possible occurrence of obstructive sleep apnoea may gradually occur. In the absence of timely intervention and treatment, pulmonary collapse (atelactasis) or recurrent pneumonia may occur; in extreme cases, respiratory difficulties may lead to death of the individual.[9,12]

As a result of the gradual changes, **complications in the musculoskeletal system** occur, causing limitations in bipedal locomotion and other motor skills (jumping, stair walking, standing upright). During early adolescence, individuals experience a complete loss of locomotor abilities, which can lead to the rapid development of muscle contractures and subsequent development of physical deformities (most commonly scoliosis of the spine). Due to the limited independent mobility, the mobility of the upper limbs is gradually restricted. Other complications within the musculoskeletal system include the development of osteoporosis as a result of standard glucocorticoid treatment. This is usually initiated around the age of five and is essential to maintain independent motor skills for as long as possible. Osteoporosis combined with progressive muscle dysfunction can lead to the occurrence of vertebral compression fractures. When any fracture

occurs, it is necessary to think about the possibility of developing fat embolism syndrome and to adapt physical activity to this.[9,13,14]

Cardiological complications in DMD are most often manifested in the form of progressive cardiomyopathy with associated arrhythmias. Dystrophin is as dysfunctional in cardiomyocytes as in skeletal muscle cells. The pathological findings of the cardiac wall are heterogeneous in nature and usually involve the left ventricle whose wall is hypertrophied. There is a combination of cardiac muscle atrophy with secondary myocardial wall remodelling. These changes occur because of the incorporation of adipose tissue with fibrotic changes, which occur through the loss of cardio myocytes. However, the term cardiomyopathy is an inaccurate term in this case, as myocardial changes in DMD patients do not meet all diagnostic criteria. In most cases, the normal dimensions of the ventricles are preserved, but their wall is always thinned and systolic function is reduced. Impaired systolic function is manifested by a reduced ability to contract the heart muscle. This issue is subsequently reflected in the clinical manifestation, in the form of symptoms of heart failure - edema, dyspnea.[9,11,15]

Complications of the immune system. During muscle regeneration in a healthy person, there is a mutual communication between the immune system and the muscle in question. The result of this cooperation is the activation of macrophages, T-lymphocytes and other cells of the immune system, which play a major role in immune processes. The innate and consequently adaptive processes of immunity normally maintain a balance between pro-inflammatory cytokines (IL-6, TNFα) and anti-inflammatory cytokines (IL-4, IL-10), thus keeping the muscle in homeostasis. In DMD patients on long-term corticosteroids, this balance is disturbed and muscle tissue damage thus overrides regenerative processes. The immune system is also responsible for collagen deposition and its disturbance leads to fibrosis, which in turn affects the condition of muscle tissue, which loses its elasticity and overall functionality. The inflammatory mechanisms in DMD depend on many modulators and result in loss of muscle mass and its replacement by adipose tissue.[16] Patients with DMD receiving long-term immunomodulatory therapy may also have a reduced immune response to vaccination, which significantly reduces their protection against serious, potentially fatal diseases.

Other complications that can occur in a person with DMD are associated with dystrophin dysfunction, which is present in the retina, central nervous system and kidneys. In this context, possible relationships with various types of cognitive and neuropsychiatric disorders or other behavioral problems should be considered. These issues have a major impact on the quality of life of the individual concerned, but also have an impact on the cooperation and outcome of rehabilitation. It is therefore always necessary to check that the patient understands the instructions and is able to perceive them correctly. This verification should be done as soon as possible so that rehabilitation and other therapies can be carried out correctly. A correct assessment of the condition can help the socialization of the patient with DMD, the integration into society according to his/her capabilities and prevent exclusion already from the childhood group.[9,17,18]

3.2 DIAGNOSTICS

The diagnosis of DMD is based on the clinical picture, which is then confirmed by genetic testing. Although it is currently an incurable disease, early diagnosis is crucial in order to slow down the rapid progression of the disease. It is essential to initiate targeted treatment from which the individuals concerned can benefit.[19] The clinical picture will serve as a marker for further action to rule out any other serious pathologies. First and foremost, laboratory collections are completed, where in particular elevated levels of transaminases (alanine aminotransferase, aspartate aminotransferase) and muscle enzymes (creatine kinase, lactate dehydrogenase and myoglobin) are detected. In this type of disease, as a rule, an increased level of creatine kinase is found.[20,21] In case of finding elevated levels of the above-mentioned transaminases and muscle enzymes, molecular genetic diagnosis is proceeded to. If the deletion or duplication of the DMD gene is confirmed, the result is considered positive. If the gene mutation is found to be positive in the child, the mother should also be subjected to genetic testing. The most commonly used quantitative method is Multiplex Ligation-dependent Probe Amplification (MLPA), which can be used to determine the extent of deletions or duplications at the level of exon resolution.[22] If the MLPA method does not show any findings, diagnosis is made by Next Generation Sequencing (NGS), which allows searching for minor changes and point mutations in the gene.[23] If inconclusive, a muscle biopsy can be performed to confirm or refute the absence of dystrophin. The diagnosis of this disease is very time consuming, usually the whole process takes about 3-6 months. If any suspicion of a possible pathology arises, ideally by the 18th month of life,

all concentration is devoted to diagnosis, which should be complete by the individual's second birthday.[20] Genetic predisposition to DMD arises from a genetic mutation in the dystrophin gene, which is located on the short arm of chromosome Xp21.2. It is an X-linked recessively inherited disease, but there are also cases in which the disease is observed to arise from new mutations.[24] Mutation of the dystrophin gene inhibits the production of the protein "dystrophin". As it is an X-linked recessively inherited disease boys are more often affected than girls. Carriers are usually asymptomatic, but there are cases where the opposite is true. If the DMD gene is translocated and disrupted, the disease can manifest in heterozygous females. A translocation is a structural rearrangement that results from a chromosomal rearrangement. If a given change occurs on the X chromosome and at the same time the protein-coding gene is disrupted, it is possible for a woman to be affected by an X-linked recessive disorder.[25]

3.3 CURRENT TREATMENT OPTIONS

DMD is a disease that currently still cannot be cured. However, a number of studies are underway that suggest a potential pathway to improving the health of the affected person or to curing DMD directly. An integral part of the care of patients with DMD is a multidisciplinary approach that includes clinical care, rehabilitation aimed at improving physical condition and respiratory function, as well as psychosocial therapy, including for family members.[26] Through targeted treatment approaches, it is possible to influence the course of the disease, both positively and negatively.

The most common pharmacological treatment used is corticosteroids, which have an anti-inflammatory effect that is used to slow the progression.[4]

The latest experimental treatment is gene therapy, the essence of which is to correct the code for dystrophin production in all neuromuscular cells. The European Medicines Agency (EMA) has decided to grant a marketing authorisation for Duvyzat. It is a histone deacetylase (HDAC) inhibitor that modulates uncontrolled HDAC activity in dystrophic muscles. HDAC inhibition can lead to a reduction in inflammation and scarring of tissue. The drug is indicated for DMD patients aged six years and older, with the requirement that the patient must be able to walk.[27] The first targeted gene therapy was the administration of the active ingredient ataluren. This causal therapy was especially crucial for children with nonsense type point mutations for DMD. Ataluren acts on ribosomes and improves their function. The nonsense mutation is characterized by a stop codon on the mRNA that causes the production of a truncated non-functional dystrophin, and ataluren, in contrast, promotes the production of a full-length functional protein (dystrophin).[28] However, in November 2024, the EMA issued a negative opinion against this active substance and the European Commission issued a final decision on 28 March 2025 not to renew the product's marketing authorisation.[29] This involves skipping exons outside the reading frame in the dystrophin transcript using antisense oligonucleotides, which are special small pieces of modified RNA, to produce a partially functional dystrophin. Four antisense oligonucleotides - eteplirsen, casimersen, golodirsen and viltolarsen - have been approved by regulatory agencies for the treatment of patients with mutations.[30,31] The first patients were dosed in 2018, but clinical trials have been halted at various stages due to the occurrence of adverse events, which include thrombotic microangiopathy induced by complement activation causing renal dysfunction, problems related to the emergence of adeno-associated viruses serving as carriers, and patient death.[30]

Endocrinological treatment is an integral part of the care of people with DMD, the aim of which is to monitor the growth and development of the individual and, if necessary, to detect hormonal insufficiency and initiate hormone replacement therapy. The mainstay of DMD treatment is the administration of glucocorticoids, which have the negative side effect of impairing linear growth that is already impaired by the disease. Regular examinations are required every 6 months until the end of growth development. Examination of linear growth impairment should include standard screening tests to evaluate endocrine hormones or other abnormalities associated with growth impairment. Here, the cooperation of the physiotherapist and the parents who are in daily contact with the person with DMD is inseparable.[13,19] Standard glucocorticoid therapy brings with it a number of negative side effects, the main side effect being an adverse impact on bone health. Bone formation is inhibited, which subsequently leads to an increased risk of fractures. Individuals with DMD are most at risk of developing vertebral compression fractures. When this complication occurs, treatment is resorted to using bisphosphonates, which bind to the surface of the bones to slow their

wear and tear. The pharmacological approach to treatment can be combined with a non-pharmacological approach, which includes physical therapy, nutritional support and the use of compensatory aids.[32]

Gastrointestinal and nutritional care for individuals with DMD is primarily focused on avoiding overweight or malnutrition. At the same time, there is a need to focus on a balanced diet with adequate intake of calories, protein, fluid, vitamins (especially vitamin D) and minerals.[33] There is currently no research relating to the nutritional needs of individuals with DMD, so these individuals are treated as the general population. As the disease progresses, problems with the swallowing mechanism itself gradually emerge. If dysphagia is present, a collaborative approach with a speech therapist and a physiotherapist experienced in orofacial rehabilitation should be taken. The goal of physiotherapy techniques is to restore swallowing function, improving the strength and range of motion of the structures involved in swallowing. Oromotor exercises, swallowing manoeuvres or phonorespiratory exercises are most commonly used.[34] Other complications that may be encountered include impaired gut motility, gastroesophageal reflux or constipation. Any complications that arise can be discussed with a gastroenterologist who will then consider the insertion of a nasogastric probe.[8]

Insufficiency of dystrophin in the heart results in cardiomyopathy; therefore, long-term **cardiologic care** is necessary in persons with DMD. Cardiovascular disease is one of the most common causes of morbidity and mortality in people with DMD.[19] Previously, it was rare if a cardiologist was part of the disciplinary team right from the start of the diagnosis. Nowadays, there is a strong emphasis on cardiology care from the confirmation of the disease, but setting up treatment is not straightforward. If signs of heart failure are detected (shortness of breath, oedema, rhythm disturbances) the individual is subjected to further investigations which are repeated annually. Based on the data obtained from the examinations performed, the physical condition and cardiorespiratory adaptation to exercise can be evaluated, which is the information needed to develop a physiotherapy plan.[35]

Orthopaedic and surgical treatment. In the early (walking) phase, orthopaedic treatment is mainly aimed at preventing contractures. The method of first choice is regular stretching and splinting. Surgical treatment is resorted to in patients with severe contractures according to the state of health. As a rule, ankle plantar flexion contracture is the first to develop, followed by knee, hip, elbow, wrist and forearm supination. If equinovarus deformities of the leg are present, extension or tenotomy of muscles such as the m. flexor hallucis longus, flexor digitorum longus and Achilles tendon is resorted to. However, corrective surgeries are not recommended for hip and knee contractures, as significant correction cannot be achieved here and may result in weakening of the hip flexors, affecting the ability to walk. The same results apply to the management of trauma.[36–38] At all stages of the disease, patients are monitored for the development of scoliosis. In the early (walking) stage, an examination of the facets using the Adams' forward bend test is usually sufficient. If the test is positive, this is followed by a radiographic examination, which can be used to accurately determine the degree of scoliosis. It is recommended that the examination revealing spinal deformities be performed annually.[37] People with DMD are at increased risk of osteopenia and osteoporosis and fractures. In particular, fractures of the lower limbs are a complication that leads to long-term immobilization and limitation of even normal activities, which negatively affects the overall condition. Conservative treatment involves fixation of the fracture, but a longer period of immobilisation and a possible risk of poor adhesion or decubitus fractures must be taken into account. With the operative approach, the immobilisation time is reduced and there is also less risk of complications.[39] Within peer reviewed studies, we find evidence of the effectiveness and positive impact of operative interventions in people with DMD, where in the sample studied with femur fracture (n=10), five patients were treated conservatively and five patients were treated operatively. The femoral fractures treated operatively in the persons with DMD healed properly and were physically the same as before surgery. In the second group studied, there was no recovery of walking ability.[39] The surgical approach gives the possibility to start verticalization and to start loading the musculoskeletal system in a very short time.[40]

Palliative treatment should be part of a multidisciplinary approach in the treatment of the disease. Currently, palliation is still understood by the professional community to be associated with cancer, but DMD/BMD is an incurable disease that ends in death, so palliative treatment has its place. There is a need to raise awareness of palliative services among the general public and the professional community, even in

the early stages of the disease. The family should be informed about the possibilities of using palliative treatment when needed.[41]

3.4 REHABILITATION IN DUCHENNE MUSCULAR DYSTROPHY

Rehabilitation offers many options and approaches, but the key to achieving the goals set for a person with DMD lies in a multidisciplinary approach that includes care in physiotherapy, occupational therapy and speech therapy with overlap into clinical disciplines. The goals of rehabilitation are to improve the quality of life or at least maintain the existing condition. Rehabilitation is always determined individually according to the desired goals and symptoms of the individual. It is necessary to emphasize regularity, systematicity and early initiation of rehabilitation from the confirmation of the diagnosis. Physiotherapists and occupational therapists play a key role in the comprehensive care of people with DMD, with the main aim of their intervention being to support and maintain the patient's functional abilities within the limits set by the progression of the disease.[42–44]

The individual physical and psychological condition of people with DMD makes it impossible to develop a specific plan for a given stage of the disease, but it is possible to follow general principles. The division of the different phases according to Mayhew et al. (2025):

1. Early ambulatory phase (walking) - in the framework of which the emphasis is mainly on early intervention and maintaining muscle strength for the longest possible time;
2. late ambulatory phase (still walking but with difficulty) - the physiotherapist should prevent muscle contractures by regular stretching;
3. early non-ambulatory phase (still walking and standing, but doing most things in a wheelchair) - follows the previous phase, but here problems related to impaired mobility have already occurred; physiotherapists and occupational therapists introduce the necessary assistive devices according to the patient's functional status;
4. late non-ambulatory phase (wheelchair-bound, without ventilatory support) - the exercise units are mainly aimed at maintaining upper limb mobility and rehabilitating breathing;
5. late non-ambulatory care with ventilatory support - procedures are recommended here to maximize the individual's quality of life.[45]

Within kinesiotherapy, elements of concepts such as analytical procedures, the Vojt method, the Bobath concept, proprioceptive neuromuscular facilitation to sensorimotor stimulation can be used. However, care must be taken to avoid eccentric contraction, which is a contraindication.

Education of DMD patients by a physiotherapist is crucial so that patients understand when and how to use each muscle in daily activities and what to avoid or reduce the load (a typical example is sitting). Exercise activities must take place under conditions that do not lead to harm to the patient. This includes providing a suitable warm room, while avoiding cold minimises muscle fibre contraction and reduces the risk of injury.

A very common complication is the development of contractures and deformities, so physiotherapy emphasises prevention. It is recommended to perform muscle stretching every day according to the state of health. It starts with stretching the muscles of the ankle, knee and hip joints, as well as the muscles of the wrists and hands, and in the case of advanced conditions, the neck area is not forgotten. In the extreme positions, “stretching” must never be performed, as this could lead to damage to the muscle tissue and also to the muscle attachment. Orthotic devices such as the Ankle-Foot Orthosis (AFO) are used for passive stretching during the night or in the non-ambulatory stages of the disease. The earlier the application is started, the better it will be tolerated by the patient. Splints of the Knee-Ankle-Foot Orthosis (KAFO) type, which are used to stabilize the knee, ankle joint and foot, are widely used, especially in the late walking or non-walking stages. Splints for the wrist and hand area, which are used only in the non-ambulatory phases of the disease, have the same representation.[19,46] The previously used application of Hyasa (hyaluronidasum) or Hyasa iontophoresis in combination with several hours of positioning is now contraindicated. This form of rehabilitation damages muscle tissue in the long term and the application alone could cause serious reactions.[47,48]

Aerobic activities have an irreplaceable place, primarily used water activities or cycling (currently with the help of an electric bike). The emphasis should always be on respecting fatigue and not overloading the

patient. Hydrotherapy and hydrokinesiology open up new possibilities for physical activities that individuals with DMD would no longer be able to do in normal life. The aquatic environment helps in relieving locomotion. A thermopositive temperature is recommended for exercise. However, there are limitations for individuals with DMD, which include the difficulty of exercising in an aquatic environment, which can be partially overwhelming, but at the same time adds a constant resistance that can lead to patient overload. The use of balneotherapy is an advantageous adjunct to therapy, but it must be considered that it involves mostly passive immersion of the patient in water without active motor participation.[49,50]
Apart from the aquatic environment, which has applications in the early stages of the disease, it is also possible to use robotic assisted suspension systems that provide dynamic support for verticalization and gait training. The available studies show positive effects in the application of this therapy, including inter-individual variability in the clinical status of patients, which determines the suitability of different therapeutic modalities. In some individuals, the use of an aquatic environment is not feasible due to the presence of external osteosynthetic fixators.[40]

Published studies suggest that vibrating platforms have a high potential at a frequency of 7-24 Hz and an amplitude of 2-4 mm. The result shows that bone mass, muscle strength and some bone markers did not decrease during the study period, which is beneficial information for possible further research in this area of rehabilitation care. Adverse effects after vibration platform exercise occurred in the form of itching or burning of the legs, dizziness or pain in the jaw and cervical spine.[32,51]

A wide range of treatments can be used from physical therapy. Almost any form of thermotherapy can be applied prior to stretching contractures, including positive balneotherapy. Application of analgesic currents with the aim of pain relief.[52]

The use of more alternative approaches, as a complementary form of rehabilitation, in the form of hippotherapy, canister therapy and the use of other species of animals, has a positive impact on people with DMD not only physically, such as relaxation of spastic tissues, but also on the psychological state. The use of hippotherapy has been shown to improve balance and postural control, maintain muscle strength and possibly improve range of motion.[50,53,54]

The recent situation with COVID-19 has shown the fragility of the system and ensuring the availability of qualified care. However, studies are emerging that also suggest a lack of availability of skilled specialist care for people with DMD.[45] Modern technology has opened up the possibility of telerehabilitation, which has become particularly important during the coronavirus pandemic. Regulation lockdowns for some DMD patients may have disrupted the complexities of multidisciplinary care due to isolation. At that time, online physiotherapist-led training courses were developed that focused primarily on respiratory physiotherapy, autostretching with a particular focus on the lower and upper limbs. A set of six tests was used to assess the motor skills of the individual, while the patient was asked to submit photo documentation on a regular basis. At the end of the pandemic, 84% of patients (n=44) involved in telerehabilitation reported satisfaction with home rehabilitation and completed outpatient rehabilitation.[55] For pediatric patients, it is necessary to take into account their cognitive status to determine whether telerehabilitation is an appropriate tool for that particular child. An undeniable benefit of both synchronous and asynchronous types of telerehabilitation is improved accessibility and continuity of care, which patients with DMD/BMD require.

Social and pedagogical rehabilitation is a part of rehabilitation and serves to integrate the individual into society, to achieve the highest possible level of self-sufficiency, independence and higher education. Integrating an individual into a collective is not easy. Teachers need to be invited into the multidisciplinary team and informed about the disease, its manifestations and future progression. If the child is at a disadvantage, an assistant will need to be provided and an individual education plan established. Children with DMD are sometimes more than averagely intelligent, however, when they are integrated into a standard school, they may experience stress at the new situation and learning difficulties may arise. In the case of mainstream education, maximum comfort should be ensured in the form of adherence to the ergonomics of the working environment. Socialisation and integration into a peer group of classmates has an impact on the psyche and a significant influence on the progression of the disease. For a child with DMD, the limiting factor for the choice of school is also its readiness and accessibility.[18,56]

4 CONCLUSIONS.

The paper summarizes the current knowledge about Duchenne muscular dystrophy, including its etiology, clinical picture, diagnosis and treatment options. It demonstrates the importance of a multidisciplinary approach where rehabilitation plays a key role in maintaining quality of life. Emphasizes an individualized approach to each patient with respect to disease progression. It also describes modern therapeutic methods, including experimental gene therapy and the importance of telerehabilitation. It highlights the common complications of the disease and the need for early recognition. The evolution of care for patients with DMD is dynamic and further improvements in treatment outcomes can be expected with the advent of new technologies. A historical review reveals the complexities of earlier diagnosis and limited therapeutic options. Although it is still an incurable disease, current approaches allow to significantly influence the course of the disease. In the future, gene therapy is expected to be a major milestone that will lead to a change in the health of DMD/BMD individuals.

Comprehensive, early and targeted intervention remains the key to improving the prognosis of DMD patients.

5 CONFLICT OF INTREST

The authors claim to have no centre of interest.

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7 ZUSAMMENFASSUNG

Ausgangspunkt: Trotz moderner Technologie und Behandlung ist die Duchenne-Muskeldystrophie eine Krankheit, die schon in jungen Jahren zum körperlichen Abbau des Körpers und sogar zum Tod führt.
Gruppe: Ziel dieses Artikels ist es, die Ergebnisse der Entwicklung der Behandlung und Rehabilitation von Patienten mit Duchenne-Muskeldystrophie darzustellen und die derzeitigen Rehabilitationsmöglichkeiten zu definieren, um so lange wie möglich einen Zustand der Unabhängigkeit und Selbstversorgung zu erhalten.
Methoden: Die Suche erfolgte in erster Linie auf der Grundlage festgelegter Schlüsselwörter.
Ergebnisse: Die Rehabilitation bei DMD muss auf einem umfassenden Ansatz beruhen. Der Erfolg der Physiotherapie hängt von einem frühen Beginn, einem individuellen Ansatz, Kontinuität und einer engen interdisziplinären Zusammenarbeit ab. Neben dem Bewegungsapparat müssen auch die orofazialen und respiratorischen Funktionen, das Herz-Kreislauf-System und die psychosoziale Stabilität des Patienten und seiner Familie angesprochen werden. Neben der Verwendung von Elementen aus klassischen Therapiekonzepten, der Balneologie und der Hydrokinesiologie erscheint die Einbeziehung der Telerehabilitation sinnvoll, um die Zugänglichkeit und Kontinuität der Versorgung zu gewährleisten.
Schlussfolgerungen: Der historische Rückblick zeigt die Komplexität der früheren Diagnose und die begrenzten therapeutischen Möglichkeiten. Obwohl es sich nach wie vor um eine unheilbare Krankheit handelt, können die derzeitigen klinischen und rehabilitativen Ansätze den Verlauf der Krankheit erheblich beeinflussen.

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