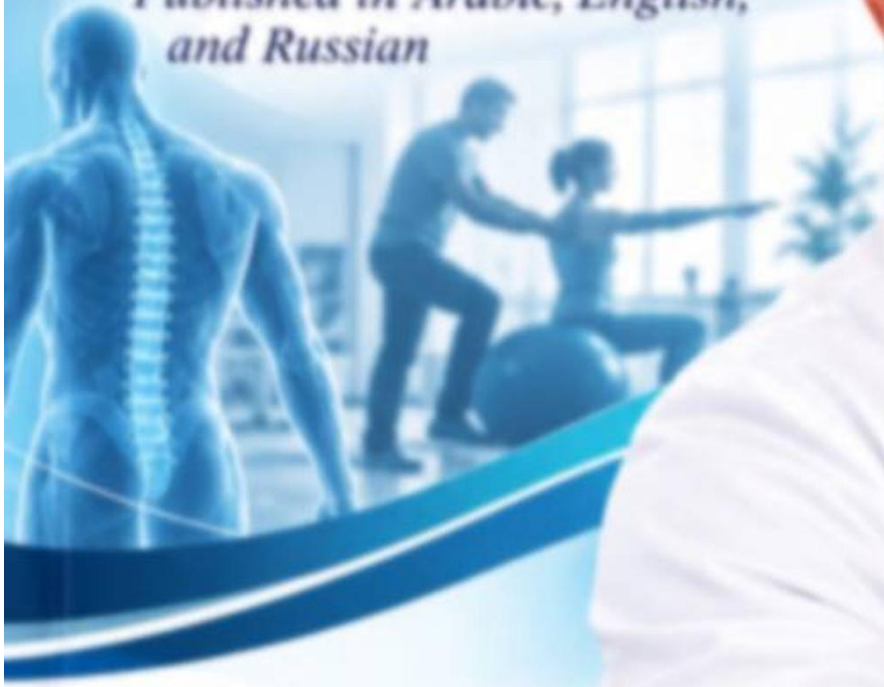
Scientific and Educational Series

36.6 – 366 Days a Year

PHYSICAL THERAPY FOR GENETIC DISORDERS

An International medical textbook
for medical colleges, higher medical
education institutions,
and practicing physicians

Published in Arabic, English, and Russian





Author:
Scientific Doctor
Vladimir Maletskyi

36 Disorders

6 Thematic Parts

Владимир Малецкий

Physical Therapy for Genetic Disorders

«Автор»
2026

Малецкий В. В.

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A genetic diagnosis names the cause. It does not name the limits.Physical Therapy for Genetic Disorders is a 36-chapter clinical textbook on physical therapy and medical rehabilitation for patients with hereditary disorders. It bridges the gap between genetic diagnosis and clinical rehabilitation practice, treating each condition as a clinical challenge that calls for an individualized strategy.Six parts cover hereditary muscle diseases, disorders of the nervous system, genetic syndromes, skeletal disorders, connective tissue and multisystem syndromes, and metabolic and lysosomal storage diseases.Every chapter follows the same ten sections, from genetics and clinical presentation to therapeutic exercise, orthoses, contraindications, and recommendations for families.For students of medical colleges and universities, physical therapists, rehabilitation physicians, neurologists, orthopedists, pediatricians, and medical geneticists.

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Владимир Малецкий
Physical Therapy for Genetic Disorders

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Medical Disclaimer

This textbook is intended for medical education, scientific reference, and professional development.

The information presented is not a substitute for individualized clinical assessment, professional medical judgment, or applicable clinical guidelines.

Physical therapy and rehabilitation interventions must be selected according to the patient's diagnosis, clinical condition, functional status, contraindications, and individual needs.

Medical knowledge and clinical recommendations evolve over time. Readers should consult current scientific evidence, relevant clinical guidelines, and applicable professional standards before implementing any diagnostic or therapeutic approach.

The author and publisher do not guarantee specific clinical outcomes from the application of the methods discussed in this publication. Clinical decisions and patient care remain the responsibility of appropriately qualified healthcare professionals.

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Abstract

Physical Therapy for Genetic Disorders is a specialized medical textbook by scientific doctor Vladimir Maletskyi, devoted to contemporary approaches to physical therapy, medical rehabilitation, and functional support for patients with hereditary disorders.

The publication is part of the authors scientific and educational series 36.6 — 366 Days a Year, united by the idea of continuous care for human health, the advancement of medical knowledge, and the improvement of clinical practice.

The textbook is based on a systematic study of 36 hereditary and genetically determined disorders grouped into six thematic parts.

Each of the 36 chapters addresses an individual disorder or a group of related genetic disorders and follows a uniform ten-section framework, progressing from an understanding of genetic mechanisms and clinical manifestations to the development of an individualized physical therapy program, prevention of complications, and recommendations for the patient.

The textbooks principal distinguishing feature is its combination of fundamental medical knowledge with practical principles of rehabilitation.

Disorders are considered not only as genetic diagnoses but also in terms of their effects on movement, muscle strength, coordination, independence, learning ability, social adaptation, and quality of life.

Structure of the Textbook

Part I. Hereditary Muscle Diseases

The first part is devoted to disorders involving primary damage to muscle tissue, progressive muscle weakness, contractures, and impaired motor function.

It covers Duchenne and Becker muscular dystrophies, limb-girdle muscular dystrophies (Erb–Roth), facioscapulohumeral muscular dystrophy (Landouzy–Dejerine), myotonic dystrophy (Steinert), and Emery–Dreifuss muscular dystrophy.

Particular attention is paid to maintaining motor activity, appropriate exercise dosing, prevention of contractures, orthopedic support, and monitoring of cardiac and respiratory complications.

Part II. Hereditary Disorders of the Nervous System

The second part examines hereditary neurological disorders involving motor neurons, peripheral nerves, neural pathways, and structures of the central nervous system.

It includes spinal muscular atrophy, Charcot–Marie–Tooth disease, hereditary spastic paraplegia, Friedreich ataxia, ataxia-telangiectasia (Louis–Bar syndrome), and Huntington disease.

The main areas of physical therapy include maintaining functional mobility and independence, correcting gait and balance impairments, reducing the functional consequences of spasticity and ataxia, preventing falls, and helping patients adapt to progressive neurological limitations.

Part III. Genetic Syndromes

The third part addresses genetic syndromes in which motor development disorders may coexist with cognitive, behavioral, sensory, and systemic manifestations.

It examines Down, Joubert, Rett, Angelman, and Prader–Willi syndromes, as well as fragile X syndrome.

Special importance is given to early intervention, development of motor skills, support for communication, promotion of independence, creation of a safe environment, and interdisciplinary care for patients of different ages.

Part IV. Hereditary Skeletal Disorders

The fourth part is devoted to disorders associated with congenital abnormalities in bone and cartilage formation, skeletal deformities, joint instability, and orthopedic complications.

It includes achondroplasia, osteogenesis imperfecta, diastrophic dysplasia, campomelic dysplasia, Larsen syndrome, and congenital spondyloepiphyseal dysplasia.

Topics include safe physical activity, prevention of fractures and secondary deformities, orthotic management, functional adaptation, and specific considerations for rehabilitation in patients with increased bone fragility or spinal instability.

Part V. Hereditary Connective Tissue Disorders and Multisystem Syndromes

The fifth part brings together disorders in which abnormalities of connective tissue, the musculoskeletal system, and internal organs require special consideration when prescribing physical exercise.

It examines Marfan syndrome, Ehlers–Danlos syndromes, Bethlem myopathy, tuberous sclerosis, neurofibromatosis type 1, and Wilson disease.

The emphasis is on assessing individual risks, preventing injuries to joints and soft tissues, addressing cardiac and neurological complications, and combining motor rehabilitation with specialized medical follow-up.

Part VI. Hereditary Metabolic Disorders and Lysosomal Storage Diseases

The final part addresses hereditary metabolic and lysosomal disorders that can cause progressive involvement of muscles, the nervous system, the skeleton, and internal organs.

It examines Pompe disease, Hurler syndrome, Hunter syndrome, Gaucher disease, metachromatic leukodystrophy, and Niemann–Pick disease type C.

Particular attention is paid to combining modern disease-specific treatments with medical rehabilitation, preserving functional independence, preventing respiratory and orthopedic complications, and maintaining quality of life in progressive disease.

Uniform Framework for Each Chapter

Each chapter of the textbook is organized into ten consecutive sections:

1. General characteristics of the disorder — genetic causes, inheritance pattern, pathogenesis, prevalence, and principal features of the disorder.

2. Clinical presentation — characteristic symptoms, age of onset, functional impairments, and potential complications.

3. Goals of medical rehabilitation — identification of individual objectives, functional priorities, and expected outcomes.

4. Therapeutic exercise — principles of designing an exercise program, dosing of physical loads, and development and preservation of motor skills.

5. Massage and device-based physiotherapy — potential symptomatic methods, their limitations, safety considerations, and assessment of clinical appropriateness.

6. Orthopedic and functional rehabilitation — prevention of deformities and contractures, use of orthoses and assistive devices, and adaptation of everyday activities.

7. Rehabilitation for systemic and coexisting disorders — specific considerations for patients with involvement of the heart, respiratory system, nervous system, and other organ systems.

8. Contraindications and limitations — conditions and clinical risks requiring modification or temporary suspension of rehabilitation interventions.

9. Recommendations for patients and relatives — organization of home-based assistance, prevention of complications, support for independence, and training in safe caregiving.

10. Conclusion — key clinical and practical findings that determine the rehabilitation strategy for the specific disorder.

This structure enables a consistent progression from theoretical understanding of a disorder to practical decisions needed in the day-to-day work of medical professionals.

International Educational Format

The textbook is intended to be available in three language versions: Arabic, English, and Russian.

The Arabic, English, and Russian editions are designed to broaden access to professional knowledge and to apply a common educational concept across different countries.

Intended Readership

The publication is recommended as educational and reference literature for students at medical schools and colleges, students at higher medical education institutions, and specialists in physical therapy and medical rehabilitation.

The textbook is also of practical interest to rehabilitation physicians, neurologists, orthopedists, pediatricians, medical geneticists, physiotherapists, and other professionals involved in the comprehensive care of patients with hereditary disorders.

The material can be used for independent professional study, preparation of thematic classes, discussion of clinical cases, and planning of individualized rehabilitation programs.

Concept of the Publication

36.6 is more than the title of a medical series. It symbolizes the continuity of medical knowledge and care for human health throughout all 366 days of the year. The series 36.6 — 366 Days a Year embodies a clinical credo: the symbol of normal body temperature and the number of days in a year, brought together in a single title, remind us that caring for health is an everyday commitment rather than a one-time course of treatment.

The patient is at the center of this textbook: not only the diagnosis, but also the persons ability to move, communicate, learn, work, and retain the greatest possible degree of independence.

A genetic diagnosis identifies the cause of a disorder. Medical rehabilitation helps identify a persons capabilities.

This is the principle to which scientific doctor Vladimir Maletskyis book Physical Therapy for Genetic Disorders is dedicated.

Why This Book Is a Significant Contribution

First. This is a textbook in which genetics and physical therapy are treated not as parallel disciplines but as a unified clinical approach. Genetic disorders cease to be viewed as a sentence and instead become conditions to which rehabilitation technologies can be applied.

Second. The book was written by a scientific doctor with substantial international experience working with patients with genetic disorders. Behind every chapter are clinical experience, knowledge of the evidence base, and an understanding of what actually works in clinical practice and what remains theoretical.

Third. The series 36.6 — 366 Days a Year is a philosophy that places the patient, rather than the diagnosis, at its center. Thirty-six disease entities, 366 days a year, and normal body temperature are three meanings that remind us that rehabilitation accompanies a person throughout the year, not only during treatment. The book teaches systematic thinking, patience, and respect for movement.

Fourth. The trilingual publication serves as a bridge between medical schools. Arabic, English, and Russian span regions with different healthcare systems but a shared challenge: hereditary disorders require rehabilitation, while professionals trained to provide it remain in short supply.

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Chapter 1. Duchenne Muscular Dystrophy

Part I. Hereditary Muscle Diseases
Duchenne Muscular Dystrophy (DMD)

1. General Characteristics of the Disease

Duchenne muscular dystrophy is a severe inherited neuromuscular disease characterized by progressive weakness and degeneration of skeletal muscles, as well as involvement of the cardiac and respiratory muscles.

The disease is caused by pathogenic variants in the DMD gene, located on the X chromosome. This gene is responsible for the production of dystrophin, a protein that maintains the stability of muscle cell membranes. In its absence or in cases of severe deficiency, muscle fibers are damaged during contractions and are gradually replaced by adipose and connective tissue.

The disease occurs predominantly in boys, with an incidence of approximately 1 case per 3,500–5,000 live-born boys (on average, approximately 1 in 5,000). In rare cases, girls who carry a DMD gene variant may also experience muscle weakness and signs of cardiac involvement.

The first signs usually appear between 2 and 5 years of age. Despite the progressive nature of the condition, timely treatment and comprehensive rehabilitation can help patients retain functional abilities for longer.

2. Clinical Presentation

Early manifestations include delayed motor development and difficulty running, jumping, and climbing stairs. The child falls frequently and has difficulty rising from the floor. Gowers sign is characteristic: the patient rises by successively using the hands to push against the legs.

Pseudohypertrophy of the calf muscles, pelvic girdle weakness, and a characteristic waddling gait are observed.

As the disease progresses, joint contractures, postural abnormalities, scoliosis, and loss of independent ambulation develop. Later, the risk of respiratory failure and dilated cardiomyopathy increases.

The diagnosis is confirmed by molecular genetic testing. Blood creatine kinase activity is usually markedly elevated.

3. Goals of Medical Rehabilitation

The primary goal of rehabilitation is to preserve existing functional abilities for as long as possible without causing additional muscle damage.

Key priorities include preventing contractures, maintaining range of motion, supporting independence, preventing orthopedic complications, and helping the patient adapt to everyday life.

The program must take into account the patients age, disease stage, motor activity, and cardiovascular and respiratory status.

4. Therapeutic Exercise

Therapeutic exercise is the principal method of motor rehabilitation in Duchenne muscular dystrophy.

Three stages are conventionally distinguished: early ambulatory (independent walking and preserved upper-limb function), late ambulatory (assisted or limited walking and increasing contractures), and nonambulatory (wheelchair mobility). The goals and intensity of therapeutic exercise differ at each stage.

In the early stages, moderate aerobic exercise and gentle active and active-assisted movements are recommended. Aquatic exercises may be beneficial provided the patients condition is stable.

Particular attention is given to maintaining mobility of the ankle, knee, and hip joints.

Shortened muscles should be stretched slowly, without pain or excessive force. Session duration and intensity are determined individually.

Training to the point of muscular failure is strongly discouraged. High-intensity resistance exercise, jumping, exercises with a substantial eccentric component, and excessive resistance should be avoided.

Pain, prolonged fatigue, or deterioration in motor abilities after exercise requires a reduction in the workload and reassessment of the patients condition.

5. Massage and Device-Based Physical Therapy

Massage may be used as an adjunctive procedure to improve patient comfort, reduce the subjective sensation of muscle tension, and facilitate care.

Gentle, superficial techniques are preferable. Deep kneading, intense vibration, and painful manipulation of weakened muscles are undesirable.

It is important to understand that massage does not restore dystrophin or prevent the destruction of muscle fibers.

Electrical stimulation (TENS and NMES), therapeutic ultrasound, infrared therapy, and magnetic therapy lack convincing evidence that they slow the progression of Duchenne muscular dystrophy. TENS may be considered for selected pain-management purposes, but not as a treatment for the dystrophy itself. Routine use of NMES to strengthen affected muscles is not recommended without specialist assessment.

Rehabilitation should be based primarily on therapeutic movement, prevention of contractures, orthopedic support, and respiratory care.

6. Orthopedic Rehabilitation

As the disease progresses, muscle imbalances and restrictions in joint mobility develop.

Ankle-foot orthoses, including those intended for nighttime use, may be prescribed according to individual indications. Appropriate limb positioning and regular assessment of the spine are important.

After the loss of independent walking, the patient should be provided with a suitable wheelchair, trunk support, and equipment for safe transfers.

Particular attention should be paid to preventing falls and fractures. Patients receiving long-term glucocorticosteroid therapy are at increased risk of reduced bone mineral density.

7. Respiratory Rehabilitation

As respiratory muscles weaken, cough effectiveness and the ability to maintain adequate lung ventilation decrease.

Retention of bronchial secretions, nocturnal hypoventilation, and recurrent respiratory infections may occur.

Patients require regular assessment of respiratory function. Assisted coughing techniques and noninvasive ventilation are used when indicated.

Respiratory rehabilitation is conducted in collaboration with a pulmonologist and specialists in neuromuscular diseases.

8. Contraindications and Limitations

When planning procedures, progressive muscle weakness, cardiomyopathy, reduced bone strength, and possible respiratory impairment must be considered. Patients with low bone mineral density and respiratory muscle weakness require particular attention, since even relatively minor trauma may cause a fracture, while excessive physical exertion may increase respiratory fatigue.

The following should be avoided:

Intensive strength training and exercise to exhaustion.

Aggressive stretching of muscles and joints.

High-impact exercises associated with an increased risk of falling.

Deep, painful massage of weakened muscles.

Physical exertion during acute infection, fever, or unstable cardiopulmonary status.

Routine use of device-based procedures without justified indications.

If severe shortness of breath, chest pain, dizziness, or sudden marked weakness occurs, the session must be stopped and a medical assessment performed.

9. Recommendations for Family Members

The family plays a key role in maintaining the patients motor activity and quality of life.

Parents are taught how to perform home exercises safely, position the limbs, use orthoses, and prevent falls.

It is important not to force the child to exercise through pain or pronounced fatigue.

As the disease progresses, the living environment must be adapted to ensure access to education, communication, and everyday activities.

Psychological support for the patient and family members should accompany rehabilitation at all stages of the disease.

10. Conclusion

Duchenne muscular dystrophy requires comprehensive multidisciplinary management involving a neurologist, rehabilitation physician, cardiologist, pulmonologist, orthopedist, and other specialists.

Modern treatment may include glucocorticosteroids and certain mutation-specific therapies for patients who meet the established indications.

Medical rehabilitation does not eliminate the genetic defect; however, it helps preserve motor function, prevent secondary complications, and maintain the greatest possible degree of independence.

The fundamental principle guiding rehabilitation professionals in Duchenne muscular dystrophy is to preserve movement without overloading weakened muscles.

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Chapter 2. Becker Muscular Dystrophy

Part I. Hereditary Muscle Diseases
Becker Muscular Dystrophy (BMD)

1. General Characteristics of the Disease

Becker muscular dystrophy is a hereditary neuromuscular disease characterized by progressive weakness, predominantly of the proximal skeletal muscle groups, and possible involvement of the heart muscle.

The disease is associated with pathogenic variants in the DMD gene located on the X chromosome. Unlike Duchenne muscular dystrophy (see Part I, Chapter 1), in which functional dystrophin is virtually absent, Becker muscular dystrophy usually involves the continued partial production of an altered or shortened protein. Consequently, the disease progresses more slowly in most cases. However, the severity of clinical manifestations varies considerably, ranging from moderate muscle weakness to severe motor limitations and cardiomyopathy.

The condition occurs predominantly in males. Some women who carry pathogenic variants in the DMD gene may also develop muscular and cardiac manifestations. The incidence of Becker muscular dystrophy is approximately 1 case per 18,000–19,000 live-born boys (according to different estimates, from 1:18,450 to 1:19,000). The disease is considerably less common than Duchenne muscular dystrophy.

2. Clinical Presentation

The first symptoms may appear in childhood, adolescence, or adulthood.

The most characteristic difficulties involve running, climbing stairs, and rising from a seated position. Weakness of the pelvic girdle and thigh muscles gradually develops.

Patients may exhibit calf muscle pseudohypertrophy, a waddling gait, Gowers sign, muscle pain, and increased fatigability.

Over time, joint contractures, postural abnormalities, and a reduced ability to walk independently may occur.

An important feature of Becker muscular dystrophy is the possibility of significant myocardial involvement even when muscle strength is relatively preserved. In some patients, cardiomyopathy becomes the predominant clinical manifestation.

The diagnosis is confirmed by molecular genetic testing. Creatine kinase activity is usually elevated.

3. Goals of Medical Rehabilitation

The primary goal of rehabilitation is to maintain the patients functional independence while minimizing the risk of additional muscle damage.

The rehabilitation program aims to preserve joint mobility, prevent contractures, maintain achievable levels of muscle activity, improve balance, and prevent falls.

Unlike more severe forms of dystrophinopathy, many patients with Becker muscular dystrophy retain the ability to walk independently for a prolonged period.

Adapted physical activity and independence in occupational and daily activities therefore play an important role.

Cardiovascular assessment is required before prescribing exercise.

4. Therapeutic Exercise

Therapeutic exercise should be prescribed individually, taking into account muscle strength, fatigability, and cardiac status.

When motor function is preserved, moderate aerobic activities may be used, including walking on level ground, exercising on a stationary bicycle with low resistance, swimming, and water-based exercise.

Cautious low-resistance exercises aimed at maintaining muscle strength may be permitted if they are well tolerated and approved by a specialist.

Gentle stretching of the calf muscles, hip flexors, and other muscle groups prone to shortening is recommended.

Exercises to improve balance, coordination, and stability during walking are beneficial.

Patients must not be pushed to marked muscular exhaustion. Excessive resistance training, intensive eccentric exercise, and exercising through pain should be avoided.

The duration, frequency, and intensity of exercise are selected individually. Muscle pain, weakness, and recovery time should be assessed after exercise.

5. Massage and Device-Based Physical Therapy

In Becker muscular dystrophy, massage may be included in a rehabilitation program when localized muscle discomfort or associated pain is present.

Particular attention should be paid to gentle treatment of overloaded muscle groups, taking into account the degree of weakness and individual tolerance of the procedure.

Gentle techniques without substantial pressure are preferred. Painful kneading, intense vibration, and aggressive manipulation of weakened muscles are undesirable.

Massage does not correct the genetic defect or stop the progression of muscular dystrophy.

TENS may be used for symptomatic pain relief in selected cases; however, disease-specific evidence of effectiveness in Becker muscular dystrophy is insufficient.

The use of NMES should be considered in light of residual muscle function, the degree of fatigability, and the patients functional goals. When selecting stimulation parameters, the specialist must take into account the characteristics of the dystrophic process and the individuals tolerance of the load.

In Becker muscular dystrophy, therapeutic ultrasound, infrared therapy, and magnetic therapy may be considered by a specialist as adjunctive methods for associated symptoms when there are independent indications. Their inclusion in the program depends on individual tolerance, safety, and the anticipated symptomatic benefit; evidence that these procedures influence the progression of dystrophy is insufficient.

6. Orthopedic Rehabilitation

As muscle weakness progresses, the likelihood of restricted joint mobility, gait abnormalities, and contracture development increases.

Regular assessment of the feet, knee and hip joints, spine, and the patients stability is necessary.

Orthoses, walking aids, and devices that facilitate daily activities may be used according to individual indications.

In cases of pronounced weakness, falls should be prevented and the surrounding environment adapted.

When prescribing orthoses, their potential effects on preserved motor function must be considered.

7. Respiratory and Cardiac Rehabilitation

Respiratory disturbances in Becker muscular dystrophy generally develop later and do not occur in all patients. Nevertheless, respiratory function should be monitored as muscle weakness progresses.

Cardiac monitoring is particularly important.

Cardiac follow-up should begin at diagnosis and include electrocardiography (ECG), echocardiography, and, when necessary, cardiac magnetic resonance imaging (MRI). According to GeneReviews recommendations, cardiac assessment is performed annually, or more frequently if myocardial dysfunction is identified, as determined by the cardiologist. If cardiomyopathy develops, medications may be prescribed to treat heart failure and prevent complications. In such cases, physical activity should be coordinated with the cardiologist. Specialized respiratory support methods are used in cases of respiratory failure.

8. Contraindications and Limitations

Rehabilitation planning must take into account the possible combination of muscle weakness, cardiac arrhythmias, and reduced exercise tolerance. Particular caution is required in the presence of cardiomyopathy and arrhythmias. Even when muscle strength is relatively preserved, physical activity may need to be restricted by the cardiologist following cardiac assessment.

The following should be avoided:

High-intensity resistance training and exercise to exhaustion.

Substantial eccentric loading.

Aggressive stretching and painful massage techniques.

Physical exertion in the presence of unstable cardiomyopathy or significant arrhythmias.

Exercise during acute infections, fever, or marked deterioration in general condition.

Device-based procedures without justified indications and a safety assessment.

If chest pain, fainting, severe shortness of breath, or palpitations occur, the activity must be stopped and a medical assessment arranged.

Severe muscle pain, a sudden increase in weakness, or dark urine after exercise requires urgent evaluation to rule out muscle injury.

9. Recommendations for Patients and Family Members

Patients and their families should understand that moderate, regular activity is preferable to either complete avoidance of movement or excessive training.

A home program may include gentle exercises, safe walking, maintenance of joint mobility, and rest periods.

Adequate recovery time after exertion should be observed, and prolonged overexertion should be avoided.

Working-age patients may require workplace adaptations, changes in the nature of physical demands, and ergonomic aids.

Particular attention should be paid to regular cardiac follow-up, even when muscle weakness is mild.

10. Conclusion

Becker muscular dystrophy is characterized by considerable clinical variability and often progresses more slowly than Duchenne muscular dystrophy (see Part I, Chapter 1).

Timely medical rehabilitation helps maintain motor activity, prevent contractures, and preserve independence.

Cardiac monitoring is particularly important because the severity of cardiac involvement does not always correspond to the degree of muscle weakness.

Comprehensive care should involve a neurologist, a rehabilitation physician, a cardiologist, a physical therapist, and other specialists.

The central principle of rehabilitation in Becker muscular dystrophy is to maintain physical activity while balancing the functional benefits of exercise against the safety of the muscular and cardiovascular systems.

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Chapter 3. Limb-Girdle Muscular Dystrophies (Erb–Roth)

Part I. Hereditary Muscle Diseases
Limb-Girdle Muscular Dystrophies (LGMD)

1. General Characteristics of the Disease Group

Limb-girdle muscular dystrophies are a group of hereditary neuromuscular disorders characterized by progressive weakness primarily affecting the muscles of the shoulder and pelvic girdles.

Historically, some limb-girdle forms were referred to as Erb–Roth muscular dystrophy. Modern classification is based on specific genetic abnormalities.

Numerous LGMD subtypes are known, associated with variants in CAPN3, DYSF, SGCA, FKRP, and other genes. Inheritance may be autosomal recessive or autosomal dominant. Age at symptom onset and rate of progression vary considerably. According to Orphanet, the combined prevalence of limb-girdle muscular dystrophies is estimated at approximately 1 in 44,000–123,000 people. The frequency of individual genetic subtypes varies substantially between populations.

2. Clinical Presentation

The disease may manifest in childhood, adolescence, or adulthood.

The most characteristic difficulties involve climbing stairs, rising from a chair, running, and walking long distances. When the shoulder girdle is affected, patients may have difficulty raising their arms, dressing, and performing everyday activities.

A waddling gait, Gowers sign, scapular winging, muscle atrophy, and joint contractures may be observed. Certain genetic forms may involve the myocardium and respiratory muscles.

Diagnosis includes neurological examination, creatine kinase testing, muscle imaging, and molecular genetic testing. Identification of the subtype is necessary for appropriate planning of monitoring and rehabilitation.

3. Goals of Medical Rehabilitation

The rehabilitation strategy for LGMD is determined primarily by the genetic subtype and the distribution of muscle weakness. For some patients, the priority is maintaining stable walking; for others, it is preserving the ability to raise their arms, dress independently, and perform daily activities. Functional goals are therefore established after assessing shoulder- and pelvic-girdle function and the patients overall condition.

Accordingly, the main goals of medical rehabilitation include:

Preserving mobility of the shoulder and hip joints and preventing contractures;

Maintaining muscular endurance within individual tolerance;

Improving balance and stability during walking;

Preserving upper-limb function and independence in daily activities;

Preventing falls.

4. Therapeutic Exercise

Therapeutic exercise is the foundation of the rehabilitation program.

When independent mobility is preserved, the specialist may include stationary cycling with minimal resistance, graded walking, and pool-based exercise. Particular attention is given to pelvic-girdle stability, trunk control, and preservation of upper-limb function. Exercise must take into account possible asymmetry of muscle weakness and differences between genetic subtypes of the disease.

Exercises for the shoulder- and pelvic-girdle muscles, maintenance of joint range of motion, and balance training are important.

Stretching should be performed gently and without pain. Excessive eccentric loading, training to exhaustion, and exercises causing marked muscle soreness should be avoided.

After each session, fatigue, muscle pain, and recovery of motor function are assessed.

5. Massage and Physical Therapy Modalities

In limb-girdle muscular dystrophies, the choice of massage technique depends on the distribution of muscle weakness and the condition of the shoulder and pelvic girdles. The specialist assesses tenderness, restricted movement, and compensatory overuse of individual muscle groups before deciding whether gentle manual techniques are appropriate.

Gentle techniques without intensive kneading or painful pressure on weakened muscles are preferred.

TENS may be used for selected pain syndromes but does not affect the genetic mechanism of the disease.

The potential use of NMES depends on the genetic subtype of LGMD and the pattern of muscle involvement. The method requires careful selection of parameters and assessment of the patients response, because evidence of its effectiveness in different forms of limb-girdle dystrophy is limited.

Therapeutic ultrasound and infrared therapy may be considered adjunctive physical therapy modalities when concomitant pain syndromes and muscle discomfort are present. Their use is primarily directed toward symptomatic goals, improving patient comfort, and increasing tolerance of rehabilitation interventions.

Magnetotherapy (PEMF) may also be considered an adjunct within a comprehensive rehabilitation program. The specialist makes an individual decision based on the patients clinical condition, potential contraindications, and the limited scientific evidence for the effectiveness of this method specifically in LGMD.

A rational combination of therapeutic exercise, massage, and additional device-based modalities allows the rehabilitation program to be individualized according to the patients functional capabilities. The choice of each intervention should be based on specific therapeutic goals and its anticipated clinical benefit.

6. Orthopedic Rehabilitation

As the disease progresses, joint contractures, postural abnormalities, gait instability, and reduced mobility may develop.

Regular assessment of joint mobility and gait analysis are necessary.

The choice of orthopedic aids in LGMD depends on whether pelvic- or shoulder-girdle muscle involvement predominates. In patients with lower-limb weakness, the specialist selects orthoses and walking aids to compensate for gait impairment and reduce the risk of falls. With marked shoulder-girdle involvement, the patients ability to perform manual tasks independently and use adaptive devices for daily living must be assessed. If mobility is substantially reduced, an individually fitted wheelchair and positioning aids are considered.

Particular attention should be given to fall prevention and maintaining safe activity.

7. Respiratory and Cardiac Rehabilitation

Some genetic subtypes of LGMD are associated with cardiomyopathy, cardiac rhythm disturbances, and respiratory muscle weakness.

Cardiac and respiratory monitoring is therefore determined according to the identified genetic variant.

When indicated, assessments include ECG, echocardiography, pulmonary function testing, and other examinations.

When cough effectiveness decreases, respiratory support techniques may be used; in respiratory failure, noninvasive ventilation may be required.

Physical activity is adjusted if cardiopulmonary complications are identified.

8. Contraindications and Limitations

Rehabilitation planning must take into account the genetic subtype, degree of muscle weakness, and risk of muscle fiber damage. In certain genetic forms of LGMD, particularly some dysferlinopathies, intense physical exertion may be accompanied by substantial muscle damage. The specialist should therefore consider the genetic diagnosis, muscle enzyme levels when indicated, and the patients response to previous sessions.

The following should be avoided:

High-intensity training to exhaustion;

Excessive eccentric loading;

Aggressive stretching and painful deep massage;

Exercises with a high risk of falling;

Exertion in the presence of unstable cardiopulmonary disorders;

Unjustified use of device-based physical therapy modalities.

The onset of sudden weakness, prolonged muscle pain, severe shortness of breath, or dark urine after physical exertion requires discontinuation of exercise and medical evaluation.

9.Recommendations for Patients and Family Members

Patients should be informed about the importance of regular but appropriately graded physical activity.

A home program may include gentle joint mobility exercises, safe walking, balance exercises, and rest periods.

Physical overload should be avoided, and the home environment should be adapted in a timely manner if independence decreases.

The specialist teaches family members safe ways to assist with rising from a chair, transferring from bed, and negotiating obstacles at home. In cases of marked shoulder-girdle weakness, particular attention is given to preserving independence in dressing, eating, and performing manual activities, using adaptive devices when necessary.

Psychological support, maintenance of social activity, and vocational adaptation are also important.

10. Conclusion

Rehabilitation in limb-girdle muscular dystrophies requires an understanding of the clinical heterogeneity of this group of disorders. Even when muscle weakness appears similar, patients may differ substantially in their rate of progression, tolerance of physical exertion, and risk of cardiorespiratory complications.

Successful specialist care is therefore determined not by the number of prescribed procedures, but by the accuracy of individual assessment, evidence-based selection of methods, and timely adjustment of the rehabilitation program.

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Chapter 4. Facioscapulohumeral Muscular Dystrophy (Landouzy–Dejerine)

Part I. Hereditary Muscle Diseases
Facioscapulohumeral Muscular Dystrophy (FSHD)

1. General Characteristics of the Disease

Facioscapulohumeral muscular dystrophy (Landouzy–Dejerine) is a hereditary disorder that primarily affects the muscles of the face, shoulder girdle, and upper limbs. As the disease progresses, the muscles of the trunk and lower limbs may also become involved.

Two main genetic forms are recognized: FSHD1 and FSHD2. Their development is associated with dysregulation of DUX4 gene expression, which causes pathological changes in muscle cells. The more common form, FSHD1, results from a contraction of D4Z4 repeats in a specific region of chromosome 4.

A characteristic feature of the disease is marked variability in clinical manifestations, even among members of the same family. In some patients, motor limitations remain moderate for decades; in others, weakness gradually spreads to additional muscle groups. According to various epidemiological estimates, the prevalence of FSHD is approximately 4–10 cases per 100,000 people. It is among the relatively common forms of hereditary muscular dystrophy, and most diagnosed cases are FSHD1.

2. Clinical Presentation

The first manifestations often occur during adolescence or early adulthood. Patients may find it difficult to whistle, close their eyes tightly, smile, or maintain facial expressions.

Examination frequently reveals scapular winging (winged scapulae), limited ability to raise the arms above shoulder level, and weakness of the muscles that stabilize the scapulae. Muscle involvement is often asymmetric.

When the abdominal and back muscles are affected, posture changes and lumbar lordosis may become more pronounced. Weakness of the ankle dorsiflexors makes walking more difficult and increases the risk of tripping.

Some patients experience chronic pain, increased fatigue, and sleep disturbances. In early-onset disease, hearing impairment and retinal vascular abnormalities may also occur.

The diagnosis is confirmed by specialized molecular genetic testing.

3. Goals of Medical Rehabilitation

In FSHD, rehabilitation focuses primarily on preserving useful movements of the shoulder girdle and the ability to perform activities independently with the hands.

The main goals include maintaining the available range of motion in the shoulder joints, improving scapular positioning and control, preventing compensatory postural abnormalities, and preserving stable gait.

Another focus is reducing the functional consequences of asymmetric weakness. The specialist assesses not only muscle strength but also the patients ability to lift objects, dress, use household items, and perform occupational tasks.

When facial muscles are involved, the adequacy of eye closure, articulation, and other functions affecting everyday life are also assessed.

4. Therapeutic Exercise

The therapeutic exercise program is developed after assessing shoulder mobility, scapular position, trunk muscle strength, and the patients stability.

For the upper limbs, active and active-assisted movements are performed within the available range of motion. Exercises with the forearms supported on a table or sliding surface help reduce the effects of gravity and facilitate arm movement.

Sessions may include exercises for scapular positioning and control, trunk stabilization, and preservation of thoracic spine mobility. It is important not to attempt to correct scapular winging mechanically through excessive resistance training.

For lower-limb weakness, balance exercises, weight-shifting activities, and safe gait training are used. Stationary cycling and pool-based exercise may be included according to the patients motor abilities.

Moderate aerobic activity and individually selected resistance exercises may benefit physical fitness. The specialist assesses not only tolerance during the session but also the preservation of movement quality afterward. Current studies indicate that individually tailored moderate resistance training in FSHD may be well tolerated and help maintain muscle function. However, study findings are heterogeneous, so exercise intensity and effectiveness must be assessed in relation to the degree of muscle involvement and the patients response.

5. Massage and Device-Based Physical Therapy

In FSHD, massage may be used to relieve associated muscle discomfort, particularly in the neck, shoulder region, and back, where compensatory overuse commonly develops.

Treatment must take into account the uneven distribution of muscle involvement. Gentle techniques aimed at relieving tenderness and relaxing overworked areas are preferred.

TENS may be considered for selected pain syndromes. The potential use of NMES is assessed cautiously, taking into account the degree of atrophy and the functional condition of the muscles to be stimulated.

Therapeutic ultrasound, infrared therapy, and magnetotherapy are not disease-specific treatments for FSHD. Their use for associated symptoms may be discussed on an individual basis, but convincing evidence of their effectiveness specifically for this dystrophy is lacking.

When selecting physical therapy modalities, priority is given to a specific functional goal: facilitating movement, reducing pain, or improving tolerance of the core exercise program.

6. Orthopedic Rehabilitation

One of the most characteristic orthopedic problems in FSHD is inadequate scapular stabilization, which limits upper-limb movement.

To assess potential functional benefit, the specialist may perform a manual scapular fixation test, observing how much the range of arm elevation increases.

In selected patients, surgical fixation of the scapula to the chest wall may be considered. This intervention requires specialist assessment because it is not suitable for everyone and may be associated with complications. A key selection criterion is preserved deltoid muscle strength: when the deltoid is markedly weak, scapular fixation does not provide functional improvement.

For ankle dorsiflexor weakness, ankle-foot orthoses are used to reduce the risk of tripping. In cases of postural abnormalities, supportive devices are selected and the workplace is adapted.

Contrary to a common assumption, orthopedic management in FSHD aims not only to prevent deformities but also to improve functional use of the upper limbs.

7. Respiratory and Cardiac Rehabilitation

In most patients, the heart muscle is not a primary site of involvement; therefore, intensive cardiac monitoring without relevant indications is generally unnecessary.

Respiratory failure also does not develop in every patient, but its likelihood increases with marked generalized weakness, postural abnormalities, and substantially reduced mobility.

If shortness of breath, morning headaches, daytime sleepiness, or signs of nocturnal hypoventilation occur, respiratory function should be assessed and the need for respiratory support evaluated.

In severe cases, noninvasive ventilation may be required. Physical activity in such cases is planned jointly with relevant specialists. If cough effectiveness declines, assisted coughing techniques and respiratory physiotherapy may be used, as in other neuromuscular diseases.

8. Contraindications and Limitations

The greatest caution in rehabilitation for patients with FSHD is required with exercises involving prolonged holding of the arms in an elevated position or substantial resistance imposed on the shoulder-girdle muscles.

Activities that cause significant pain, prolonged muscle fatigue, or deterioration in previously achievable movements are not recommended.

In the presence of substantial scapular instability, uncontrolled overhead resistance exercises should be avoided. Ankle dorsiflexor weakness increases the risk of falls when walking on uneven ground.

If eye closure is incomplete, ophthalmological assessment is required to prevent corneal injury. Respiratory impairment must also be considered when prescribing physical activity.

9.Recommendations for Patients and Families

Patients are advised to organize daily activities so as to minimize, where possible, the need to hold their arms above shoulder level for prolonged periods.

Placing frequently used items at a convenient height, using forearm supports, and adapting the workplace can make daily tasks considerably easier.

In cases of pronounced facial muscle weakness and incomplete eyelid closure, recommendations from an ophthalmologist for protecting the ocular surface should be followed.

Family members should recognize that muscle weakness that appears mild may substantially hinder certain activities. Assistance should aim to preserve the patients independence rather than completely replace their own efforts.

Periodic functional assessment allows timely adjustment of home exercises and selection of assistive devices.

10. Conclusion

Facioscapulohumeral muscular dystrophy (Landouzy–Dejerine) is distinguished by its characteristic distribution of muscle involvement, asymmetry of symptoms, and considerable variation in the rate of progression.

A central feature of rehabilitation is addressing shoulder-girdle function: preserving the ability to use the arms, compensating for scapular instability, and preventing secondary postural abnormalities.

Well-organized care makes it possible to focus on the motor functions that matter most for the individual patients independence in everyday life.

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Chapter 5. Myotonic Dystrophy (Steinert)

Part I. Hereditary Muscle Diseases
Myotonic Dystrophy Type 1 (DM1)

1. General Characteristics of the Disease

Steinert myotonic dystrophy is a hereditary multisystem disease combining progressive muscle weakness with myotonia—abnormally delayed muscle relaxation after contraction.

The disease is caused by expansion of CTG trinucleotide repeats in the DMPK gene on chromosome 19. Inheritance is autosomal dominant. Symptoms may appear earlier and become more severe in successive generations, a phenomenon known as genetic anticipation.

Congenital, childhood-onset, classic adult-onset, and mild forms are distinguished. The severity of manifestations varies considerably.

Unlike many other muscular dystrophies, Steinert disease affects not only skeletal muscles but also the heart, respiratory system, endocrine system, and central nervous system. Rehabilitation must therefore account for systemic manifestations. The prevalence of myotonic dystrophy type 1 (DM1) varies considerably across regions. According to GeneReviews, the estimated worldwide average is approximately 1 in 20,000 people; the disease is more common in certain populations.

2. Clinical Presentation

A characteristic symptom is difficulty relaxing the hand after a forceful grip. The patient cannot immediately open the fingers after a handshake or holding an object. Repeated movement sometimes reduces myotonia, producing the warm-up phenomenon.

Muscle weakness often begins in the distal limbs, particularly the finger flexors and ankle dorsiflexors. Atrophy of the forearm, hand, and lower-leg muscles gradually develops.

Facial muscle weakness, ptosis, altered facial expression, and articulation difficulties are typical. The masticatory and swallowing muscles may also be involved.

Extramuscular manifestations include cardiac conduction disturbances, arrhythmias, cataracts, insulin resistance, gastrointestinal disorders, and pronounced daytime sleepiness.

The congenital form is associated with generalized hypotonia, respiratory disturbances, and delayed motor development. Diagnosis is confirmed by molecular genetic testing for CTG-repeat expansion in the DMPK gene.

3. Goals of Medical Rehabilitation

The primary goal is to preserve practical hand function and the patient's ability to perform precise everyday movements despite the combination of muscle weakness and myotonia.

When designing a program, the specialist identifies several priorities: improving control of grasping and releasing objects, maintaining limb muscle strength, preventing falls due to ankle dorsiflexor weakness, and preserving independence in self-care.

Additional goals include improving exercise tolerance, preventing respiratory complications, and identifying swallowing difficulties promptly.

Rehabilitation goals must take account of daytime sleepiness, cognitive characteristics, and cardiac status, as these factors may substantially affect the safety and outcomes of therapy.

4. Therapeutic Exercise

Therapeutic exercise in Steinert disease has a particular focus: in addition to maintaining muscle function, it must develop the ability to control the initiation and completion of movement.

Hand exercises include working with soft objects of different shapes, sequential finger flexion and extension, transferring small objects, and training grasp coordination. Prolonged maximal gripping should be avoided because it may worsen difficulty with relaxation.

Repeated low-intensity movements sometimes alleviate myotonia through the warm-up phenomenon. This transient improvement in relaxation must be distinguished from ordinary muscle fatigue.

Lower-limb exercises address lifting the forefoot, maintaining stability while turning, and negotiating small obstacles. When functional capacity is sufficient, graded walking and stationary cycling may be used.

Structured moderate-intensity aerobic and resistance training may improve muscle function, walking endurance, and certain quality-of-life measures in patients with DM1. However, the available studies remain limited in size; therefore, the program must be individualized after assessing cardiovascular status, the severity of fatigue, and the patient's response to physical exertion.

5. Massage and Device-Based Physical Therapy

Massage is considered primarily an adjunctive measure for associated pain and muscular discomfort. Its application requires an understanding of the differences between myotonic contraction, muscle weakness, and ordinary muscle tension.

Gentle massage techniques may be used when tenderness is present. Intense mechanical stimulation and painful techniques are inadvisable, especially if they provoke prolonged muscle contraction.

TENS may be used for symptomatic pain relief when independently indicated. The potential use of NMES must be assessed individually: its effectiveness specifically for correcting myotonia or strengthening affected muscles in DM1 is not sufficiently established.

Therapeutic ultrasound, infrared therapy, and magnetotherapy likewise do not specifically target the myotonic mechanism. Their use for associated pain requires consideration of expected benefits, contraindications, and the limitations of clinical evidence.

Particular caution is required with device-based procedures in patients with pacemakers, implantable defibrillators, or significant cardiac rhythm disturbances.

6. Orthopedic Rehabilitation

The most characteristic orthopedic problem is weakness of the ankle dorsiflexors, which prevents the patient from adequately lifting the forefoot during walking.

In such cases, ankle-foot orthoses are individually selected to facilitate safer limb advancement and reduce the risk of tripping.

For hand weakness, aids with enlarged handles, lightweight household tools, and adaptive devices for writing, eating, and personal hygiene may be used.

Selection of assistive devices must account for a characteristic feature of the disease: a patient may grasp an object relatively easily but have difficulty releasing it quickly.

When mobility is substantially reduced, mobility aids and modifications to the home environment are considered.

7. Respiratory and Cardiac Rehabilitation

Cardiac conduction disturbances are among the most serious risks in Steinert disease and may occur even when muscle weakness is moderate.

Patients require regular cardiology follow-up, including an ECG at least once a year. Holter monitoring, echocardiography, and additional investigations are performed when indicated. Significant conduction disturbances may necessitate pacemaker implantation.

Respiratory disorders may result from respiratory muscle weakness, impaired regulation of breathing during sleep, and reduced cough effectiveness.

When hypoventilation is suspected, respiratory function and sleep are assessed. Noninvasive ventilation and cough-assistance techniques are used when indicated.

Swallowing difficulties require assessment of aspiration risk and involvement of a swallowing specialist.

8. Contraindications and Limitations

Rehabilitation planning must consider the risks of arrhythmias, respiratory failure, aspiration, and pronounced daytime sleepiness.

Intensive physical exercise should not be prescribed before cardiac assessment in patients with suspected conduction disturbances or clinically significant arrhythmias.

Exercises involving prolonged maximal gripping and activities that markedly worsen myotonia, pain, or weakness require particular caution.

In patients with swallowing difficulties, exercise immediately after a large meal is inadvisable, especially in the supine position.

An important medical limitation is the increased risk of complications associated with anesthesia and sedation. Before any procedure involving anesthetic agents, the anesthesiologist must be informed that the patient has DM1.

9.Recommendations for Patients and Families

Daily activities should be organized with possible delays in muscle relaxation in mind. Patients should be given sufficient time to release objects safely, change body position, and initiate walking.

When using tools and household appliances, the possibility of unexpected difficulty opening the hand must be considered. Items that do not require prolonged forceful gripping are preferable.

Family members should watch for fainting, palpitations, pronounced daytime sleepiness, choking or coughing while eating, and signs of breathing disturbances during sleep.

Patients are advised to follow the agreed schedule of cardiology examinations and monitor associated endocrine disorders.

In congenital and childhood-onset forms, early motor intervention, speech therapy, and educational support are particularly important.

10. Conclusion

Steinert myotonic dystrophy requires a particular approach because motor impairments coexist with potentially serious systemic complications.

Its fundamental distinction from other hereditary muscle diseases is the combination of progressive weakness with impaired muscle relaxation.

The rehabilitation specialist must not only maintain physical activity but also teach the patient to use the hands safely, monitor stability during walking, and account for limitations caused by cardiac and respiratory disturbances.

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Chapter 6. Emery–Dreifuss Muscular Dystrophy

Part I. Hereditary Muscle Diseases
Emery–Dreifuss Muscular Dystrophy (EDMD)

1. General Characteristics of the Disease

Emery–Dreifuss muscular dystrophy is a rare inherited neuromuscular disease characterized by a combination of early joint contractures, gradually developing muscle weakness, and cardiac involvement.

The best-known genetic forms are associated with changes in the EMD and LMNA genes, which encode proteins involved in maintaining the structure and function of the cell nucleus. X-linked, autosomal dominant, and rarer autosomal recessive inheritance patterns occur. According to GeneReviews estimates, the overall prevalence of Emery–Dreifuss muscular dystrophy is approximately 1.3–2 cases per 100,000 population. The X-linked form is less common, at approximately 0.13–0.2 cases per 100,000 people.

Clinical manifestations may begin in childhood, and restricted joint mobility sometimes becomes apparent before a decline in muscle strength. The severity of motor and cardiac abnormalities varies considerably, even among relatives with the same genetic variant.

A distinctive feature of EDMD is that relatively mild muscular symptoms do not rule out dangerous cardiac complications.

2. Clinical Presentation

The most characteristic findings are elbow contractures, shortening of the Achilles tendons, and restricted neck flexion. Patients find it difficult to straighten their arms fully, place their heels on the floor, or bend their heads forward.

Muscle weakness initially often affects the upper-arm regions of the upper limbs and the lower-leg muscles. This distribution is termed humeroperoneal. The shoulder and pelvic girdle muscles may become involved later.

As the disease progresses, gait disturbances, difficulty climbing stairs, restricted spinal movement, and postural changes develop.

Cardiac manifestations include conduction disturbances, atrial and ventricular arrhythmias, and cardiomyopathy. Dizziness, syncope, and reduced exercise tolerance may occur. Some patients develop severe arrhythmias without preceding pronounced complaints.

Diagnosis is based on the characteristic clinical presentation, cardiac evaluation, and molecular genetic testing.

3. Goals of Medical Rehabilitation

Unlike diseases in which progressive muscle weakness is the leading problem, early identification of contractures and limitation of their consequences are of primary importance in EDMD.

The main goals are:

maintaining elbow extension and functional ankle mobility;

preserving a safe range of motion in the cervical spine;

preventing gait abnormalities associated with Achilles tendon shortening;

maintaining available muscle activity without excessive loading;

preserving independence in dressing, walking, and everyday activities.

An additional goal is to organize rehabilitation with cardiac risk in mind. The physical activity plan is developed after assessment of cardiac function and the presence of rhythm disturbances.

4. Therapeutic Exercise

Therapeutic exercise in Emery–Dreifuss disease begins with an assessment of joint mobility. The specialist determines the severity of contractures, the condition of the Achilles tendons, and the nature of restrictions in neck movement.

Regular gentle range-of-motion exercises are central. For the elbows, smooth extension within tolerated limits and exercises aimed at preserving functional arm positioning are used.

When the Achilles tendons are shortened, cautious calf stretching, ankle dorsiflexion movements, and exercises with controlled heel loading are applied. Loading must not provoke pain or tendon injury.

When neck mobility is restricted, gentle active movements are performed within a safe range. Forced flexion and abrupt, forceful manual techniques are unacceptable.

After cardiac assessment, the program may include graded walking, low-resistance exercises, and aquatic activities. With marked contractures, aquatic exercise therapy enables movement under reduced axial loading.

Because evidence on optimal exercise intensity specifically in EDMD is limited, the specialist is guided by individual tolerance, movement quality, and cardiovascular status.

5. Massage and Device-Based Physical Therapy

In EDMD, massage may be used to relieve associated muscular discomfort, particularly in areas of compensatory overstrain resulting from restricted joint mobility.

Superficial and moderate soft-tissue techniques are preferred. Established structural contractures cannot be eliminated by intensive kneading or mechanical stretching.

For pain syndromes, the potential use of TENS is considered in light of the patient's cardiac status. NMES is not a primary method of correcting contractures; the appropriateness of stimulating individual muscle groups requires specialist assessment.

Infrared therapy and therapeutic ultrasound may be considered for separate symptomatic indications, but convincing evidence of their effectiveness specifically in EDMD is lacking. Magnetotherapy is assessed similarly.

Patients with implanted pacemakers or defibrillators require particular attention. Electrical stimulation and electromagnetic procedures may be used only after compatibility with the implanted device has been verified and the cardiologist has approved their use.

6. Orthopedic Rehabilitation

Orthopedic care in EDMD is directed primarily toward managing contractures, which may substantially limit motor function even when muscle strength is preserved.

In the early stages, individually fitted orthoses may be considered to maintain functional foot positioning and preserve the available range of motion.

Surgical correction may be considered for marked Achilles tendon shortening. Decisions take account of muscle status, gait, the degree of deformity, and the potential effect of surgery on the patient's stability.

Elbow contractures require separate assessment because restricted extension interferes with everyday activities. Orthotic management and surgery do not always provide lasting restoration of mobility.

With spinal rigidity and restricted neck movement, posture should be monitored and comfortable positions selected for work, rest, and sleep.

Assessment of cardiac and anesthetic risks is mandatory before orthopedic surgery.

7. Respiratory and Cardiac Rehabilitation

Cardiac involvement in EDMD poses an independent clinical threat and does not always correlate with the severity of muscle weakness.

According to GeneReviews, evaluation should include an ECG, Holter monitoring, and echocardiography at least once a year. If conduction disturbances or cardiomyopathy are identified, monitoring should be more frequent.

Some patients require implantation of a pacemaker or an implantable cardioverter-defibrillator. In certain genetic forms, particularly those associated with LMNA, the risk of dangerous ventricular arrhythmias persists even after implantation of a conventional pacemaker.

Clearance for physical training must therefore take account of the cardiologist's assessment and, when necessary, that of a cardiac electrophysiologist.

Respiratory function may deteriorate because of respiratory muscle weakness and restricted chest mobility. When findings are normal, pulmonary function testing is recommended every two to three years; when abnormalities are present, monitoring should occur at least annually.

In respiratory failure, specialized respiratory support methods are used, including noninvasive ventilation and assistance with ineffective coughing.

8. Contraindications and Limitations

The principal limitation in EDMD is the risk of serious cardiac complications. Intensive exercise is not prescribed to patients with unevaluated arrhythmias, syncope, or suspected cardiomyopathy until specialist assessment has been completed.

Abrupt manipulations, forced elbow extension, and excessive stretching of the Achilles tendons are unacceptable when managing contractures.

Marked cervical spine rigidity requires particular caution when transferring the patient, positioning them on a treatment table, and performing physical therapy procedures.

If palpitations, presyncope, chest pain, or unusual shortness of breath occurs, the session must be stopped immediately, followed by medical assessment.

Implanted cardiac devices must be considered when selecting device-based procedures. Cardiac assessment and individualized anesthetic planning are required before surgery.

9.Recommendations for Patients and Families

Patients should understand that contractures in EDMD may develop long before a substantial loss of muscle strength. Regular assessment of joint mobility is therefore necessary even when independent walking is preserved.

Home exercises must follow specialist recommendations and be performed without forced stretching. Particular attention should be paid to foot positioning, the ability to extend the elbows, and comfort during neck movements.

Family members are advised to watch for changes in gait, toe walking, difficulty dressing, and restricted head rotation.

Recognizing signs of possible cardiac involvement is equally important. Syncope, sudden palpitations, and unexplained reduction in tolerance of usual activity require medical evaluation.

Because the disease is inherited, relatives may be offered genetic counseling and, when indicated, cardiac screening.

10. Conclusion

Emery–Dreifuss muscular dystrophy is distinguished by the characteristic combination of early contractures, humeroperoneal muscle weakness, and potentially dangerous cardiac abnormalities.

A practical feature of rehabilitation is the need to begin preventing movement restrictions before pronounced muscle atrophy develops. At the same time, cardiac status determines the permissible limits of physical activity.

Successful care requires coordinated work by rehabilitation specialists, neurologists, orthopedists, and cardiologists, because preserving joint mobility and preventing cardiac complications are complementary but distinct goals.

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Chapter 7. Spinal Muscular Atrophy (SMA)

Part II. Hereditary Disorders of the Nervous System
Spinal Muscular Atrophy (SMA)

1. General Characteristics of the Disease

Spinal muscular atrophy is a hereditary neuromuscular disease characterized by damage to the motor neurons of the anterior horns of the spinal cord and the progressive development of muscle weakness and atrophy.

The most common form is 5q-SMA, caused by biallelic pathogenic variants in the SMN1 gene on chromosome 5. Inheritance is predominantly autosomal recessive. The SMN2 gene partially compensates for the deficiency of SMN protein; its copy number influences disease severity, although it cannot precisely predict the course in an individual patient.

According to various epidemiological estimates, the incidence of 5q-SMA is approximately one case per 6,000–12,000 live births. Rates may vary depending on the population studied.

Traditionally, five clinical types are distinguished, ranging from prenatal onset (type 0) to adult onset (type IV). However, modern disease-modifying therapies alter the natural course of the disease, making the patient's actual motor status increasingly important when planning rehabilitation.

2. Clinical Presentation

The principal manifestation of SMA is symmetrical muscle weakness, usually more pronounced in the proximal parts of the limbs. Reduced muscle tone, diminished tendon reflexes, and restricted motor activity are characteristic.

In type I SMA, an untreated child does not achieve independent sitting; marked hypotonia, feeding difficulties, and respiratory disturbances develop early.

In type II SMA, the patient can sit independently but usually does not achieve independent walking. Scoliosis, contractures, and chest deformities frequently develop.

In type III SMA, independent walking is initially preserved, but difficulties with standing up, climbing stairs, and walking for prolonged periods may emerge over time.

Type IV SMA usually manifests in adulthood and is characterized by relatively slow progression.

Sensation is generally preserved. The diagnosis is confirmed by molecular genetic testing of SMN1, with determination of SMN2 copy number.

3. Goals of Medical Rehabilitation

The rehabilitation program for SMA is determined less by the formal disease type than by the patient's current motor abilities.

The main objectives include:

maintaining acquired motor skills and facilitating the development of new ones during disease-modifying treatment;

preventing contractures, spinal deformities, and complications of prolonged sitting;

preserving upper-limb function;

improving head control, trunk stability, and balance;

ensuring safe mobility and the greatest possible independence;

preventing respiratory complications.

Objective assessment of changes in the patient's condition is particularly important. Specialized scales, including CHOP INTEND, HFMSE, and RULM, are used for this purpose; the choice depends on the patient's age and motor status.

Rehabilitation outcomes are assessed through changes in specific skills, not solely through increases in muscle strength.

4. Therapeutic Exercise

Therapeutic exercise for SMA is organized around three functional categories: patients unable to sit independently, those able to sit, and those able to walk.

For non-sitters, emphasis is placed on appropriate positioning, head and trunk support, preservation of joint mobility, and creating opportunities for independent activity within the patient's capabilities. Gentle passive and active-assisted movements and safe changes of body position are used.

For sitters, the program includes seated balance exercises, pelvic positioning control, functional arm movements, and weight shifting. Standing frames and adapted standing support devices are used when indicated.

For ambulant patients, training includes rising to stand, graded walking, balance exercises, and negotiating small obstacles. Moderate aerobic activity is permissible when adequately tolerated.

Children receiving disease-modifying treatment warrant particular attention: improvements in motor abilities require reassessment of previous functional limitations and the setting of new rehabilitation goals.

Exercise intensity must match residual motor abilities and must not cause pronounced exhaustion.

5. Massage and Device-Based Physical Therapy

Massage in SMA may be used to reduce associated discomfort, facilitate care, and improve tolerance of positioning.

For patients with pronounced muscle atrophy, predominantly gentle techniques are used. Intensive kneading of weakened muscles does not restore motor neurons and should not be regarded as a method of increasing muscle mass.

TENS may be considered for certain associated pain syndromes, such as those related to prolonged sitting or orthopedic complications.

NMES requires particular caution. Because SMA involves lower motor neuron damage and denervation of muscle fibers, the effectiveness of standard neuromuscular electrical stimulation protocols may be limited.

Therapeutic ultrasound, infrared therapy, and magnetotherapy have no proven effect on the course of SMA. Their use for independent symptomatic indications must not replace therapeutic exercise, positioning, or respiratory support.

6. Orthopedic and Functional Rehabilitation

Orthopedic management is particularly important in SMA because of the high likelihood of scoliosis, contractures, and hip joint instability.

For patients who spend long periods sitting, pelvic position, trunk symmetry, and spinal condition must be monitored.

Individually fitted wheelchairs with trunk supports, headrests, and positioning accessories can improve stability and expand opportunities for independent activity.

Orthoses, standing frames, and transfer aids are used when indicated. Surgical treatment may be considered for pronounced spinal deformities, with mandatory assessment of respiratory risks.

Functional rehabilitation includes training in the use of available arm movements, workplace adaptation, and selection of alternative interfaces for controlling devices.

An important task for the specialist is not merely to correct deformities but also to create opportunities for the patient's participation in education, communication, and daily life.

7. Rehabilitation for Systemic and Associated Impairments

The most serious associated problems in SMA involve respiratory insufficiency, ineffective coughing, and swallowing difficulties.

Weakness of the intercostal muscles restricts chest expansion. In severe forms, breathing relies to a considerable extent on the diaphragm, which may be accompanied by paradoxical chest movements.

Respiratory monitoring includes assessment of cough effectiveness, pulmonary ventilation, and signs of nocturnal hypoventilation. Mechanical cough assistance and noninvasive ventilation are used when indicated.

Swallowing disorders require assessment of feeding safety because aspiration increases the risk of infectious complications. Specialized nutritional support may be required if nutritional intake is inadequate.

Current treatment of 5q-SMA includes nusinersen, risdiplam, and gene replacement therapy with onasemnogene abeparvovec for patients who meet the established indications. These treatments alter the natural course of the disease but do not eliminate the need for long-term rehabilitation support.

8. Contraindications and Limitations

In rehabilitation of patients with SMA, the degree of muscle weakness, respiratory reserve, and severity of orthopedic impairments must be considered above all.

Exercises that cause pronounced exhaustion, deterioration of breathing, or a prolonged reduction in motor activity after the session are unacceptable.

In the presence of severe osteoporosis, spinal deformities, or joint instability, traumatic manipulations and uncontrolled axial loading should be avoided.

Standing programs require assessment of tolerance of the upright position, joint condition, and fall risk.

In cases of acute respiratory infection, pronounced breathing difficulty, or signs of hypoventilation, the exercise program must be reassessed.

Particular attention is required in patients with swallowing difficulties and an increased risk of aspiration.

9. Recommendations for Patients and Families

Family members should understand that the effectiveness of home rehabilitation is determined not by the number of exercise repetitions but by how well the exercises match the patient's functional status.

For children with pronounced weakness, it is important to change body position regularly, monitor skin condition, and ensure comfortable positioning of the head, trunk, and limbs.

Patients who can sit should be given opportunities for independent play, learning, and activities using available hand movements.

When walking ability is preserved, safe daily activity should be maintained while avoiding excessive fatigue.

The family should recognize signs of respiratory deterioration: a weakened cough, difficulty clearing secretions, unusual sleepiness, and disturbed breathing during sleep.

Patients receiving disease-modifying therapy require regular reassessment of motor abilities. The emergence of new skills calls for timely adaptation of the home program.

10. Conclusion

Spinal muscular atrophy occupies a special place among hereditary neuromuscular diseases because modern disease-modifying therapies can substantially change a patient's motor prognosis.

The rehabilitation strategy is based on a functional approach that considers the ability to sit, stand, walk, and independently perform daily activities.

The rehabilitation specialist's role is not only to prevent secondary complications but also to make timely use of new motor abilities emerging during treatment.

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Chapter 8. Charcot–Marie–Tooth Disease

Part II. Hereditary Disorders of the Nervous System
Charcot–Marie–Tooth Disease (CMT)

1. General Characteristics of the Disease

Charcot–Marie–Tooth disease is a group of hereditary disorders of the peripheral nervous system characterized by progressive impairment of motor and sensory innervation, predominantly in the distal parts of the limbs.

The pathological process may affect the myelin sheath of nerve fibers or the axons themselves. Depending on the mechanism of damage, demyelinating, axonal, and intermediate forms of CMT are distinguished.

The disease is genetically highly heterogeneous. The most common form, CMT1A, is associated with duplication of a chromosomal region containing the PMP22 gene. Other forms result from variants in MPZ, GJB1, MFN2, and many other genes. Inheritance may be autosomal dominant, autosomal recessive, or X-linked.

The prevalence of CMT has traditionally been estimated at approximately 1 in 2,500 people. According to a meta-analysis published in 2023, the pooled prevalence of CMT and related hereditary peripheral neuropathies is approximately 17.7 per 100,000 population. Differences between estimates reflect the populations studied, diagnostic criteria, and case ascertainment. This is one of the most common groups of hereditary neurological disorders.

2. Clinical Presentation

Initial symptoms often appear in childhood or adolescence, although some forms manifest considerably later.

Characteristic features include weakness of the foot and lower-leg muscles, difficulty with ankle dorsiflexion, frequent tripping, and steppage gait—a walking pattern involving exaggerated lifting of the leg to prevent the toes from catching on the ground.

Pes cavus (high-arched foot), hammertoes, and ankle instability may develop gradually. Atrophy of the lower-leg muscles sometimes gives the legs a characteristic inverted champagne bottle appearance.

Reduced superficial and vibration sensation impairs awareness of foot position, especially when walking in darkness or on uneven ground. Tendon reflexes are usually diminished.

When the upper limbs are involved, difficulties arise with buttoning clothes, writing, and manipulating small objects.

Diagnosis includes neurological examination, electrodiagnostic studies, and molecular genetic testing.

3. Goals of Medical Rehabilitation

The principal challenge in CMT rehabilitation is the need to compensate for both motor weakness and sensory loss.

The main objectives are:

improving stability during standing and walking;

compensating for weakness of the ankle dorsiflexors;

maintaining the available strength of distal limb muscles;
preventing progression of contractures and the functional consequences of foot deformities;
preserving precision of hand movements;
reducing the risks of falls, skin injuries, and joint overload.

When setting functional goals, it is important to assess not only muscle strength but also walking speed, footwear condition, fall frequency, and the patient's ability to perform daily activities.

The CMTNS and CMTES scales may be used for objective assessment, and CMTPedS may be used in children.

4. Therapeutic Exercise

Therapeutic exercise aims to maintain the functional capacity of preserved muscles, improve balance, and correct altered movement patterns.

For ankle dorsiflexor weakness, active dorsiflexion exercises with light resistance are used if the remaining muscle function permits the movement. Ankle position control and weight shifting are also trained.

Balance exercises take impaired proprioception into account. Initially, stable support and visual feedback are used, with tasks gradually becoming more challenging while maintaining safety.

Regular gentle calf stretching is used to prevent shortening of the Achilles tendon. Where deformities are already established, the range of motion is determined individually.

Hand training includes finger abduction, controlled grasping of objects of different shapes, and exercises for movement precision and coordination.

Moderate resistance and aerobic activities, including stationary cycling, swimming, and graded walking, may improve physical fitness. In patients with pronounced weakness, overloading individual muscle groups should be avoided, and recovery after exercise should be monitored.

5. Massage and Device-Based Physical Therapy

In CMT, massage may be used to relieve associated musculoskeletal pain, particularly pain arising from altered gait and redistribution of loads across the joints.

When sensation is reduced, the practitioner must control pressure intensity because the patient may not always recognize excessive mechanical force in time.

TENS may be considered for selected pain syndromes. Musculoskeletal pain must be distinguished from neuropathic pain because symptomatic interventions do not have the same effectiveness in these conditions.

The potential effectiveness of NMES depends on peripheral nerve integrity and the degree of muscle denervation. In pronounced axonal disease, standard stimulation may be of limited benefit. NMES is not an established means of restoring damaged nerve fibers in CMT.

There is no convincing evidence that therapeutic ultrasound, infrared therapy, or magnetotherapy can slow CMT progression. Their use for associated pain requires separate justification; particular caution is needed when temperature and pain sensation are impaired.

6. Orthopedic and Functional Rehabilitation

Orthopedic management in CMT has a direct impact on walking quality.

For foot drop, individually fitted ankle-foot orthoses (AFOs) improve foot positioning during the swing phase and reduce the risk of tripping.

For pes cavus and ankle instability, orthopedic footwear and insoles are selected to distribute loads more evenly. Areas of excessive pressure on the foot must be checked regularly.

Surgical correction may be considered for pronounced fixed deformities. Decisions are based on assessment of muscle balance, deformity flexibility, and the anticipated functional outcome.

For patients with hand weakness, adapted writing implements, enlarged handles, and aids that facilitate household tasks are used.

Orthopedic devices should be evaluated during movement: even a technically well-made orthosis does not always produce an optimal gait for an individual patient.

7. Rehabilitation for Systemic and Associated Impairments

In most common forms of CMT, peripheral sensorimotor neuropathy remains the principal problem. Significant involvement of the heart and respiratory muscles is not an obligatory feature of the disease.

However, certain rare genetic variants may cause respiratory weakness, sleep disturbances, vocal fold involvement, and additional neurological symptoms.

Unexplained shortness of breath, nocturnal hypoventilation, voice changes, or stridor require specialist assessment.

Neuropathic pain deserves particular attention and may present as burning, tingling, or painful sensations in the feet and hands.

Pain management must address its underlying cause. Severe neuropathic pain may require specific pharmacological treatment, whereas mechanical pain caused by foot deformity may call for adjustments to orthopedic support.

8. Contraindications and Limitations

Particular caution is required during rehabilitation in patients with pronounced impairment of pain, temperature, and proprioceptive sensation.

Intensive heat treatments, excessive pressure, and traumatic manipulations may cause tissue damage that the patient does not notice promptly.

Exercises with a high risk of falling should be avoided without appropriate support, especially when foot-position control is impaired.

In the presence of pronounced deformities, prolonged weight-bearing in unsuitable footwear is inadvisable. Chafing, redness, or tenderness requires reassessment of orthoses and adjustment of the activity regimen.

Exercises causing prolonged deterioration in muscle function must be reconsidered. When treating comorbid conditions, physicians should take into account the potential neurotoxicity of certain medications.

9.Recommendations for Patients and Families

Patients with CMT are advised to inspect their feet daily, especially when sensation is markedly reduced. Skin injuries, calluses, and areas of excessive pressure should be identified promptly.

Footwear should match the shape of the feet, provide stability, and avoid additional friction. The fit of orthoses should be checked periodically.

To improve safety at home, good lighting, removal of floor obstacles, and handholds in areas with increased fall risk are advisable.

For hand weakness, adapting household items helps preserve independence without constant assistance from family members.

Because CMT comprises a group of hereditary diseases, genetic counseling may be recommended for patients and their relatives.

10. Conclusion

Charcot–Marie–Tooth disease is characterized by the combination of distal muscle weakness, sensory impairment, and characteristic orthopedic changes in the feet.

The practical value of rehabilitation lies primarily in gait correction, timely selection of orthoses, preservation of hand function, and prevention of complications related to reduced sensation.

The rehabilitation program should take account of the specific genetic subtype, the severity of axonal damage, and the patient's functional abilities. Coordinated work among a neurologist, rehabilitation specialist, orthopedist, and orthotist is particularly important.

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Chapter 9. Hereditary Spastic Paraplegia

Part II. Hereditary Disorders of the Nervous System
Hereditary Spastic Paraplegia (HSP)

1. General Characteristics of the Disease

Hereditary spastic paraplegia is a genetically heterogeneous group of neurological disorders characterized primarily by involvement of the long motor pathways of the spinal cord and the gradual development of lower-limb spasticity.

The pathological process mainly affects the corticospinal tracts, disrupting central regulation of muscle tone and voluntary movements.

Uncomplicated (pure) and complicated forms of HSP are distinguished. The former manifest predominantly as spastic paraparesis, whereas the latter may be accompanied by additional neurological impairments.

The disease is associated with numerous genetic variants, including alterations in SPAST (SPG4), ATL1 (SPG3A), REEP1 (SPG31), and SPG7. Autosomal dominant, autosomal recessive, and X-linked inheritance patterns occur.

According to epidemiological modeling, the combined prevalence of hereditary spastic paraplegias is estimated at approximately 3.6 cases per 100,000 population. Prevalence estimates for individual subtypes, including SPG4, vary according to the population studied and the methodology used.

2. Clinical Presentation

The main manifestation of the disease is gradually increasing stiffness of the lower limbs, accompanied by increased muscle tone and difficulty walking.

Neurological examination reveals exaggerated tendon reflexes, pathological plantar responses, and varying degrees of muscle weakness.

Gait becomes stiff and slow, often with insufficient knee flexion. Marked spasticity of the hip adductor muscles may lead to a crossing, or scissoring, gait.

Patients report rapid fatigue when walking, painful muscle spasms, and difficulty turning and negotiating obstacles.

Uncomplicated forms may involve impaired vibration sensation and urinary disturbances. Complicated variants may additionally feature ataxia, peripheral neuropathy, cognitive impairment, or other neurological manifestations.

Diagnosis includes neurological examination, MRI of the brain and spinal cord to exclude other causes of spastic paraparesis, and molecular genetic testing.

3. Goals of Medical Rehabilitation

The principal goal of rehabilitation in HSP is to reduce the functional consequences of spasticity while preserving sufficient muscle activity for standing and walking.

The main objectives are:

maintaining the available range of motion in the hip, knee, and ankle joints;

preventing contractures and abnormal limb postures;

improving stability and coordination during walking;

reducing painful spasms;

preserving the ability to stand up, move independently, and perform everyday activities;

reducing the risk of falls.

When setting goals, it is important to recognize that reducing muscle tone does not always improve motor function. In some patients, increased tone partly compensates for muscle weakness and helps maintain standing.

Objective assessment uses the Modified Ashworth Scale (MAS), Spastic Paraplegia Rating Scale (SPRS), 10-Meter Walk Test, and Timed Up and Go (TUG).

4. Therapeutic Exercise

Therapeutic exercise in HSP aims to maintain joint mobility, train functional movements, and reduce the impact of spasticity on gait.

For the lower limbs, slow, controlled stretching is used for muscles prone to increased tone: the hip adductors, knee flexors, and calf muscles. Stretching is performed without jerking or painful resistance.

Exercises focusing on pelvic position control, weight shifting, and stability when changing direction are particularly important.

Gait training includes practicing step length, foot placement, and safe turning. Handrails, walkers, or partial body-weight support systems are used when necessary.

When motor function is preserved, exercises are used to strengthen the hip extensors, trunk stabilizers, and other functionally important muscle groups. Stationary cycling and graded aerobic activity may be used to maintain endurance.

The program must be adapted to the degree of spasticity and muscle weakness. Prolonged fatigue, increased spasms, or worsening gait after sessions require adjustment of the workload.

5. Massage and Device-Based Physical Therapy

Massage in HSP is considered an adjunctive method for relieving muscle discomfort and painful spasms, particularly in the calves and thighs.

Gentle techniques that do not trigger a reflex increase in muscle tone are preferred. Intensive kneading and rapid stretching of spastic muscles may provoke an undesirable motor response.

TENS may be used for selected pain syndromes. NMES or functional electrical stimulation (FES) is considered individually, particularly when ankle dorsiflexion is inadequate during walking.

Electrical stimulation does not eliminate the genetic cause of the disease; any potential benefit is assessed through changes in a specific function, such as walking safety and speed.

Therapeutic ultrasound, infrared therapy, and magnetotherapy have no proven ability to slow HSP progression. Their symptomatic use requires separate indications and assessment of clinical benefit.

6. Orthopedic and Functional Rehabilitation

The most common orthopedic problems in HSP are limited ankle dorsiflexion, shortening of the calf muscles, and the development of lower-limb contractures.

Ankle-foot orthoses (AFOs) may be used when foot positioning is impaired during the swing phase of gait. Their design is selected according to the balance between spasticity, muscle weakness, and knee stability.

For substantial walking limitations, canes, walkers, and other mobility aids are used. When mobility is severely reduced, wheelchair provision is considered.

Functional rehabilitation includes training in rising from a chair, turning safely, negotiating thresholds, and transferring between different surfaces.

Established contractures may require orthopedic correction. Decisions about surgery are made after assessing muscle balance, gait characteristics, and the possible impact of the operation on the patient's independence.

7. Rehabilitation for Systemic and Associated Impairments

In complicated forms of HSP, particularly those associated with SPG11, swallowing function should be assessed specifically: clinically significant dysphagia occurs in a substantial proportion of these patients and requires timely assessment by a speech and swallowing specialist and nutritional modification.

Bladder dysfunction is a characteristic associated problem in HSP, manifesting as frequent urination, urgency, or urinary incontinence.

These symptoms require urological assessment. Medication and an individualized bladder-management program are used when indicated.

Baclofen, tizanidine, and other antispastic medications may be used to reduce marked spasticity. Botulinum toxin injections may be considered for focal increases in muscle tone, and intrathecal baclofen for severe generalized spasticity.

Pharmacological reduction of tone requires functional monitoring because excessive muscle relaxation may impair stability.

In complicated HSP, rehabilitation must also take account of ataxia, peripheral neuropathy, speech disorders, and cognitive impairment.

8. Contraindications and Limitations

When rehabilitating patients with HSP, abrupt passive movements and forced stretching of spastic muscles should be avoided.

Exercises performed without support require particular caution in patients with severe balance impairment or a tendency to fall.

Before increasing physical activity, muscle strength, the severity of contractures, and the presence of pain syndromes should be assessed.

Reducing spasticity must not become an end in itself. After botulinum toxin administration or changes in antispastic treatment, the patient's ability to stand and walk must be reassessed.

New, rapidly progressive weakness, substantial sensory disturbance, or sudden worsening of urinary function requires further investigation to exclude another neurological disorder.

9. Recommendations for Patients and Families

Patients are advised to maintain regular physical activity, alternating joint-mobility exercises with functional-skills training.

Particular attention should be paid to safe movement on uneven surfaces, stairs, and in confined spaces.

With marked spasticity, prolonged positioning that encourages the fixation of abnormal limb postures should be avoided.

Family members should help patients retain motor independence rather than routinely taking over activities they can perform themselves.

The treating physician should be informed if frequent urination, painful muscle spasms, or a substantial change in gait occurs.

Genetic counseling is recommended for patients and their relatives, taking account of the identified genetic variant and inheritance pattern.

10. Conclusion

Hereditary spastic paraplegia is a group of genetic disorders in which gradually developing lower-limb spasticity is the predominant motor impairment.

The distinctive challenge of rehabilitation is maintaining a functional balance between reducing pathological muscle tone and preserving the muscle activity needed for independent mobility.

The practical effectiveness of treatment is determined not only by reduced spasticity but, above all, by preservation of safe walking, joint mobility, and the patient's ability to carry out everyday activities.

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Chapter 10. Friedreich Ataxia

Part II. Hereditary Disorders of the Nervous System
Friedreich Ataxia (FRDA)

1. General Characteristics of the Disease

Friedreich ataxia is a hereditary neurodegenerative disease characterized by progressive impairment of movement coordination and involvement of spinal cord pathways, the peripheral nervous system, and several internal organs.

In most cases, the disease is caused by a pathological expansion of GAA repeats in both copies of the FXN gene on chromosome 9. This results in a deficiency of the protein frataxin and disruption of mitochondrial processes. Inheritance is autosomal recessive.

In countries with predominantly European populations, the prevalence of Friedreich ataxia is estimated at approximately 0.5–1.8 cases per 100,000 people, depending on diagnostic criteria. Rates vary considerably between regions.

The disease usually manifests in childhood or adolescence, although later-onset forms also occur. Progression of motor impairment is often accompanied by cardiac, orthopedic, and endocrine complications.

2. Clinical Presentation

The most characteristic manifestation is progressive ataxia—impaired coordination of movements accompanied by instability when standing and walking.

Gait becomes broad-based and irregular, with side-to-side swaying of the trunk. The patient experiences difficulty turning, changing walking speed, and performing precise motor tasks.

Damage to the posterior columns of the spinal cord and sensory nerve fibers reduces vibration sense and proprioception. As a result, the patient is less able to determine limb position without visual feedback.

As the disease progresses, dysmetria, intention tremor, dysarthria, and reduced precision of hand movements develop. Tendon reflexes in the lower limbs are often absent, although pathological pyramidal signs may persist.

Scoliosis and foot deformities, including pes cavus, are characteristic. Hypertrophic cardiomyopathy, arrhythmias, hearing and swallowing disorders, and disturbances of glucose metabolism may occur.

The diagnosis is confirmed by molecular genetic testing of FXN, including mandatory assessment of GAA repeat expansion.

3. Goals of Medical Rehabilitation

The rehabilitation strategy aims to preserve coordination, functional mobility, and patient independence while simultaneously monitoring systemic complications.

The main objectives include:

improving balance and the precision of voluntary movements;

maintaining the ability to stand, walk, and change body position safely;

preserving upper-limb function;

preventing falls and injuries;

preventing secondary contractures and restrictions of mobility;

adapting everyday activities to progressive ataxia;

maintaining endurance with due regard for cardiovascular status.

Objective monitoring uses the Scale for the Assessment and Rating of Ataxia (SARA), the modified Friedreich Ataxia Rating Scale (mFARS), and functional tests of gait and balance.

Assessment should cover not only the severity of ataxia but also the patient's ability to eat, dress, move about, and use objects independently.

4. Therapeutic Exercise

Motor rehabilitation is based on exercises for coordination, dynamic balance, and trunk postural control.

In the early stages, movements requiring a specified degree of accuracy are used: touching marked targets with the hand or foot, controlled limb movements, and sequential performance of multicomponent motor tasks.

Balance training involves gradual changes in the base of support, direction of movement, and position of the center of gravity. In pronounced sensory ataxia, the patient's reliance on visual feedback for stability must be considered.

For ambulatory patients, exercises include varying step length, negotiating small obstacles, making safe turns, and walking with external visual cues. The level of assistance is determined by fall risk.

When independent walking is lost, rehabilitation focuses on seated postural control, weight shifting, transfers between surfaces, and preservation of functional hand movements.

Low-resistance exercises and graded aerobic activity may be included after cardiac function has been assessed. Session duration is determined by tolerance and movement quality rather than the number of repetitions.

5. Massage and Device-Based Physical Therapy

Massage in Friedreich ataxia may be used to relieve secondary muscular discomfort arising from altered gait, prolonged sitting, or orthopedic deformities.

When proprioception is impaired, massage does not restore the lost mechanism of coordination and must not replace specialized motor training.

TENS may be considered for associated pain syndromes after assessing skin sensation and contraindications.

Neuromuscular electrical stimulation (NMES) is not a standard treatment for ataxia. Its use for specific motor impairments requires individualized functional justification.

Active coordination exercises, balance training, and adaptation of motor skills are of greatest importance in rehabilitation. There is no convincing evidence that therapeutic ultrasound, infrared therapy, or magnetotherapy alters the course of Friedreich ataxia.

6. Orthopedic and Functional Rehabilitation

Scoliosis and foot deformities are common orthopedic complications of Friedreich ataxia.

During growth, the spine should be assessed regularly, since progressive curvature can impair trunk stability, seated posture, and respiratory function.

Individually fitted orthoses and footwear are selected for foot deformities and ankle instability. Their purpose is to provide mechanical support without imposing additional restrictions on movement.

Patients with progressive instability may require walkers and, later, a wheelchair with trunk support and an appropriate seating system.

Functional rehabilitation includes adapting writing instruments, eating utensils, and household devices to accommodate dysmetria and tremor.

When surgical correction of scoliosis is considered, comprehensive preoperative assessment is required, especially of cardiac and respiratory function.

7. Rehabilitation for Systemic and Associated Impairments

Cardiac complications are particularly important in Friedreich ataxia. Hypertrophic cardiomyopathy and rhythm disturbances may limit exercise tolerance.

Cardiology follow-up with ECG and echocardiography is recommended at diagnosis and at least once annually. Cardiac symptoms warrant an additional, unscheduled assessment.

Because of the risk of impaired glucose metabolism, blood glucose and HbA1c should be monitored regularly. If diabetes mellitus is diagnosed, the physical activity program should be coordinated with an endocrinologist.

Dysarthria requires speech and language therapy aimed at improving speech intelligibility and using compensatory communication strategies.

Swallowing disorders are widespread in Friedreich ataxia. In one study, symptoms of dysphagia were reported in 98% of the patients examined. Silent aspiration, which occurs without a protective cough, is particularly dangerous. Swallowing function should be assessed regularly, even in the absence of pronounced complaints. When aspiration is suspected, instrumental swallowing assessment is indicated because clinical examination cannot reliably exclude it.

In later stages, possible reduction of respiratory reserve, sleep disturbances, and the need for respiratory support must be considered.

Specific pharmacological treatment is considered by the neurologist separately from the rehabilitation program, taking into account approved indications and drug availability in the country concerned.

8. Contraindications and Limitations

The main limitations relate to impaired balance, reduced deep sensation, and possible cardiac involvement.

Exercises with a high risk of falling are unacceptable without appropriate support. In pronounced ataxia, patients should avoid independently performing complex coordination tasks on unstable surfaces.

In cardiomyopathy, arrhythmias, or signs of heart failure, exercise intensity is determined following cardiological assessment.

Chest pain, fainting, marked shortness of breath, or palpitations during a session require immediate cessation of activity and medical evaluation.

In significant scoliosis and foot deformities, traumatic manipulations and exercises that cause excessive mechanical loading should be avoided.

9. Recommendations for Patients and Families

A home rehabilitation program should include regular, safe coordination exercises and maintenance of motor activity within the patient's capabilities.

Fall risk at home should be reduced by ensuring good lighting, using non-slip surfaces, and providing stable handholds where needed.

When independent walking becomes difficult, appropriate mobility aids should be selected promptly, without waiting for repeated injuries.

Family members should watch for changes in speech, choking or coughing during meals, increased fatigue, and cardiovascular symptoms.

Regular follow-up with a neurologist, cardiologist, orthopedist, and other specialists allows timely modification of the rehabilitation program.

Genetic counseling is recommended for the patient and family, taking into account the autosomal recessive inheritance pattern.

10. Conclusion

Friedreich ataxia is a hereditary neurodegenerative disease in which impaired coordination is accompanied by damage to sensory pathways and systemic complications.

The distinguishing feature of medical rehabilitation is the need to combine coordination and balance training with orthopedic support and regular cardiac monitoring.

The principal measure of rehabilitation effectiveness is preservation of the patient's functional independence, safe mobility, and ability to participate in everyday life.

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Chapter 11. Ataxia-Telangiectasia (Louis–Bar Syndrome)

Part II. Hereditary Disorders of the Nervous System
Ataxia-Telangiectasia (A-T)

1. General Characteristics of the Disease

Ataxia-telangiectasia is a rare hereditary multisystem disease characterized by progressive neurological impairment, immunodeficiency, increased sensitivity to ionizing radiation, and a predisposition to malignant neoplasms.

The disease is caused by biallelic pathogenic variants in the ATM gene on chromosome 11. The protein encoded by this gene is involved in detecting DNA damage and regulating the cellular response to it. Inheritance is autosomal recessive.

According to published epidemiological estimates, the disease occurs in approximately one in 40,000–100,000 people; lower rates are reported in some populations.

A distinction is made between the classic form, with early onset and marked progression of neurological impairment, and variant forms with milder or atypical courses.

2. Clinical Presentation

The first signs of classic ataxia-telangiectasia usually appear in early childhood, when a child begins to stand and walk independently.

Characteristic findings include an unsteady gait, impaired trunk coordination, difficulty with precise hand movements, and progressively worsening ataxia. As the disease advances, dysarthria, dystonia, choreiform movements, and peripheral neuropathy may develop.

Telangiectasias—dilated superficial blood vessels often visible on the conjunctiva and skin—are of particular diagnostic importance. They may appear after the first neurological symptoms.

Some patients exhibit oculomotor apraxia and difficulty fixing or shifting their gaze. These disturbances complicate motor tasks requiring visual guidance.

Immune abnormalities manifest as increased susceptibility to infections, particularly of the respiratory tract. Chronic bronchopulmonary complications, nutritional difficulties, and swallowing disorders may occur.

Diagnosis is based on clinical findings, immunological assessment, measurement of alpha-fetoprotein (AFP), and molecular genetic testing of ATM. Elevated AFP is characteristic but is not sufficient as a stand-alone diagnostic criterion.

3. Goals of Medical Rehabilitation

The rehabilitation program aims to preserve the patient's functional abilities while taking immune and respiratory risks into account.

The main objectives are:
maintaining head and trunk postural control;
improving stability and coordination of movements within the patient's abilities;
preserving independent mobility and self-care skills;
preventing contractures and secondary orthopedic complications;
reducing the risk of falls;
supporting effective breathing and airway clearance;

preventing functional consequences of swallowing disorders.

Outcome assessment should consider not only the severity of ataxia but also tolerance of everyday activities, frequency of respiratory complications, and the amount of assistance required.

Ataxia scales, including SARA, may be used to monitor coordination, but their results should be supplemented by an assessment of the child's actual motor abilities.

4. Therapeutic Exercise

Motor rehabilitation in ataxia-telangiectasia focuses primarily on trunk control, functional coordination, and safe performance of everyday movements.

In early stages, play-based exercises involving changes in body position, controlled weight shifting, and reaching for objects are used. When oculomotor coordination is impaired, tasks are adapted so that excessive visual demands do not interfere with movement.

For ambulatory patients, training includes stable turns, changes in stepping direction, and walking short routes with the necessary support.

As motor impairment progresses, emphasis shifts to seated stability, independent transfers, preservation of upper-limb function, and prevention of contractures.

Gentle joint mobility exercises are combined with active movement within the patient's capabilities. Prolonged intensive sessions are inappropriate in the presence of marked fatigue, respiratory symptoms, or general deterioration.

The choice of rehabilitation setting should account for the increased risk of infection and the need to limit contact with people showing signs of contagious disease.

5. Massage and Device-Based Physical Therapy

Massage may be used to relieve associated muscular discomfort and improve tolerance of positioning, but it has no proven effect on the neurodegenerative process.

In patients with pronounced weakness, deformities, or low body weight, gentle techniques without traumatic pressure are preferred.

TENS may be considered for certain pain syndromes but is not a treatment for the underlying neurological disorder.

Muscle electrical stimulation has no established efficacy against progressive ataxia and should be used only for separate functional indications.

Therapeutic ultrasound, infrared therapy, and magnetotherapy have no proven ability to alter the course of the disease. Before prescribing any device-based procedure, its purpose, safety, and associated complications must be assessed.

Physical therapy procedures must be distinguished from diagnostic irradiation: the particular radiosensitivity of these patients is primarily associated with ionizing radiation.

6. Orthopedic and Functional Rehabilitation

Progressive impairment of trunk control can contribute to scoliosis, abnormal limb postures, and difficulty sitting for prolonged periods.

Individually adapted seating systems, orthopedic supports, and positioning aids are used when needed. Ankle-foot orthoses are selected according to gait characteristics and associated muscle weakness. Their use should facilitate mobility rather than further restrict available movements.

As fall risk increases, a wheelchair may become necessary. Timely provision helps the patient continue participating in education, communication, and everyday activities.

When hand movement precision is impaired, adapted cutlery, writing implements, and assistive devices are used.

Decisions about surgical correction of severe orthopedic deformities are made after comprehensive assessment of respiratory function, immune status, and potential anesthetic risks.

7. Rehabilitation for Systemic and Associated Impairments

One of the most serious problems in ataxia-telangiectasia is the combination of immunodeficiency and chronic respiratory disease.

Recurrent infections, impaired cough mechanisms, and aspiration may lead to bronchiectasis and declining lung function.

Respiratory rehabilitation includes assessment of cough effectiveness, instruction in feasible airway-clearance techniques, and selection of assisted methods for removing bronchial secretions. Specialized cough-assist devices are used when necessary.

Immunological management may include immunoglobulin replacement therapy and infection prevention according to individual indications.

Oropharyngeal dysphagia in ataxia-telangiectasia may progress and increase the risk of aspiration-related respiratory complications. Aspiration can occur without a protective cough; therefore, recurrent pneumonia or worsening respiratory function warrants targeted assessment of swallowing safety even without complaints of choking. When indicated, instrumental swallowing evaluation, speech and swallowing therapy, modification of food consistency, and nutritional support are provided.

The increased risk of malignancies, particularly lymphomas and leukemias, is especially important. Patients require specialist surveillance.

Because of impaired DNA repair, standard approaches to radiotherapy and certain anticancer drugs may cause severe toxicity and require management by specialists experienced in treating this condition.

8. Contraindications and Limitations

The key limitation in ataxia-telangiectasia is increased sensitivity to ionizing radiation.

Whenever possible, X-ray examinations and computed tomography are replaced by methods that do not use ionizing radiation, including diagnostic ultrasound and MRI. If an examination involving radiation is essential, the decision is individualized and the dose minimized.

Radiotherapy is generally contraindicated because of the risk of severe tissue damage.

In pronounced immunodeficiency, unnecessary contact with patients who have acute infections should be avoided. Active infection, fever, or respiratory failure requires reassessment of the exercise program.

Exercises with a high risk of falling must not be performed without appropriate support.

In severe swallowing disorders, exercise immediately after feeding is inappropriate if body position or exercise intensity increases aspiration risk.

9. Recommendations for Patients and Families

Families should understand that rehabilitation for ataxia-telangiectasia must take account of not only motor impairments but also the state of the immune and respiratory systems.

Home sessions should be conducted regularly using short functional tasks suited to the child's abilities.

It is important to adapt the environment promptly to prevent falls, preserve independent participation in daily activities, and ensure appropriate positioning.

Family members should watch for persistent cough, recurrent respiratory infections, changes in breathing, choking during meals, and unexplained weight loss.

Before medical investigations, healthcare professionals must be informed of the diagnosis and the patient's increased radiosensitivity.

Genetic counseling is recommended for the family in view of autosomal recessive inheritance.

10. Conclusion

Ataxia-telangiectasia is a hereditary multisystem disease in which progressive neurological impairment is accompanied by immunodeficiency, respiratory complications, and an increased cancer risk.

The rehabilitation strategy must take account of motor development, feeding safety, respiratory function, and limitations associated with radiosensitivity.

The main objective of medical rehabilitation is to preserve available functions, prevent complications, and enable the greatest possible participation in everyday life.

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Chapter 12. Huntington Disease

Part II. Hereditary Disorders of the Nervous System
Huntington Disease (HD)

1. General Characteristics of the Disease

Huntington disease is a hereditary, progressive neurodegenerative disorder characterized by a combination of motor, cognitive, and psychiatric disturbances.

The disease is caused by a pathological expansion of CAG repeats in the HTT gene on chromosome 4. This genetic alteration leads to the production of abnormal huntingtin protein, which disrupts neuronal function, predominantly in the basal ganglia and cerebral cortex.

Inheritance is autosomal dominant. If one parent carries a pathogenic gene variant, each child has a 50% chance of inheriting it.

The prevalence of Huntington disease varies substantially across populations. Epidemiological rates are heterogeneous across European countries, as confirmed by systematic reviews. According to a 2025 meta-analysis, the pooled prevalence in sub-Saharan Africa is approximately 2.8 cases per 100,000 people.

Symptoms usually begin between the ages of 30 and 50, although juvenile forms with onset in childhood or adolescence also occur. The disease is progressive and gradually limits the patient's independence.

2. Clinical Presentation

The best-known motor manifestation of Huntington disease is chorea: involuntary, irregular, rapid movements of the limbs, face, and trunk.

In the early stages, these movements may appear as minor motor clumsiness or restlessness. Later, disturbances of gait, coordination, balance, and the precision of voluntary movements develop.

Difficulties initiating and sequencing motor actions, slowed reactions, and impaired postural control are characteristic.

As the disease progresses, dystonia, bradykinesia, and muscle rigidity may develop. In juvenile Huntington disease, rigidity and slowness of movement often predominate over chorea.

Cognitive impairment manifests as reduced ability to plan, concentrate, and perform sequential tasks. Depression, irritability, anxiety, apathy, and behavioral changes may occur.

In advanced stages, severe impairments of speech, swallowing, nutrition, and self-care develop.

The diagnosis is confirmed by molecular genetic testing of HTT with determination of the number of CAG repeats.

3. Goals of Medical Rehabilitation

The primary goal of rehabilitation in Huntington disease is to preserve functional independence for as long as possible despite combined motor and cognitive deficits.

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