

THE ROLE OF PHYSIATRY IN THE MANAGEMENT OF NEUROMUSCULAR DISEASE

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This edition of *Physical Medicine and Rehabilitation Clinics of North America* is intended to provide the reader with a comprehensive overview of the diagnostic approach, clinical characteristics, and care and management of patients with neuromuscular disease (NMD), with emphasis on eight of the most common: Duchenne muscular dystrophy (DMD), Becker muscular dystrophy (BMD), facioscapulohumeral muscular dystrophy (FSHD), limb girdle syndrome (LGS), myotonic muscular dystrophy (MMD), spinal muscular atrophy (SMA), amyotrophic lateral sclerosis (ALS), and the several types of hereditary motor sensory neuropathy (HMSN), frequently known as Charcot-Marie-Tooth (CMT) disease.

Although currently *incurable*, NMDs are not *untreatable*. These diseases are not rare, and most physiatrists will encounter a fair number of NMD patients in the course of a general physiatric practice.^{8,9} The goals of rehabilitation in patients with NMDs are to maximize functional capacities, prolong or maintain independent function and locomotion, inhibit or prevent physical deformity, and provide access to full integration into the community with good quality of life. The comprehensive management of all of the varied clinical problems associated with NMDs is an arduous task. For this reason, the multidisciplinary approach is much more effective. It takes advantage of the expertise of many clini-

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cians, rather than placing the burden on one. Management is best carried out by a team consisting of physicians; physical, occupational, and speech therapists; social workers; vocational counselors; and psychologists, among others. Ideally, owing to the significant mobility problems associated with most NMDs, the physiatrist and all the key clinical personnel should be available at each visit. Tertiary care medical centers in larger urban areas usually can provide this type of service. This may be an independent clinic or may be sponsored by one or more of the consumer-driven organizations that sponsor research and clinical care for people with NMDs, including the Muscular Dystrophy Association (MDA), the Amyotrophic Lateral Sclerosis (ALS) Association, the Charcot-Marie-Tooth International and Association groups, and the Facio-scapulohumeral Society, among others. Although the *physiatrist* (Greek *physis* for "nature" and *iatrikos* for "healing") is well-suited to direct the rehabilitation team and to oversee a comprehensive, goal-oriented treatment plan, physiatrists are co-directors of only 18% of the MDA clinics in the United States.² Bach has previously described the many major advances physiatrists have contributed to the care of NMD patients and has pointed out the rather significant need for more physiatric involvement in the care of NMD patients.²

Governmental agencies that support research in NMDs include both the National Institute on Disability and Rehabilitation Research (NIDRR), a division of the Department of Education, and the National Institutes of Health (NIH).

Although their degree and severity can vary, the characteristics or complications of most NMDs include progressive weakness, limb contractures, spine deformity, and decreased pulmonary function; some patients suffer cardiac and intellectual impairment. Based on the World Health Organization (WHO) classification of disablement, as applied to neuromuscular diseases (Table 1), impairment may be evaluated by measurements of strength, range of motion, spine deformity, cardiac and pulmonary function, and intellectual capacity.²³ Disability may be evaluated by measures of mobility and upper extremity function, cardiopulmonary adaptations, cardiac and pulmonary complications, and psychosocial adjustment. Assessment of impairment and disability should be the foundation of a thorough *physiatric* evaluation of someone with NMD. In determining impairment and disability, comprehensive physiatric evaluation of patients with NMDs involves assessing many variables; this should be done at routine intervals or as clinically indicated. Longitudinal data profiling important clinical parameters are now available for some of the major NMDs.^{6, 7, 14, 15, 18-20}

Important clinical data that should be obtained initially on each patient include: gender, birth date, family history, date of disease (symptom) onset, disease duration, dominant limb, weight and height, cardiovascular and pulmonary symptoms and findings, presence of contractures and spine deformity, muscle strength, ambulatory status (including age at cessation of ambulation and years of wheelchair use), and any treatment interventions. When relevant, these data should be

Table 1. WORLD HEALTH ORGANIZATION (WHO) DEFINITIONS OF IMPAIRMENT, DISABILITY, AND DISADVANTAGE, BASED ON ORGAN SYSTEMS

Organ	Impairment (Usually Progressive)	Disability	Disadvantage (Handicap)
Skeletal muscle	↓ Strength & endurance	↓ Motor performance ↓ Mobility ↓ Upper extremity function ↑ Fatigue	↓ Quality of life and community integration ↓ Educational opportunities ↓ Employment opportunities
Bone & joint	Joint contractures Spine deformity	↓ Function Pain & deformity	↓ Employment opportunities
Lungs	↓ Pulmonary function	Restrictive lung disease (RLD) ↑ Fatigue	↑ Dependency & disadvantage
Heart	Cardiomyopathy Conduction defects	↓ Cardiopulmonary adaptations ↑ Fatigue	
CNS	↓ Intellectual capacity	↓ Learning ability ↓ Psychosocial adjustment	

updated at each patient visit. In large clinics data can be recorded on a standardized form and entered into a computer data base so that longitudinal clinical profiles may be generated for each patient. Data obtained from the measurements described below also can be added to this data base.

Precise measures of strength are important for evaluating clinical progression in NMD as well as assessing the efficacy of any interventions. Strength traditionally has been assessed with manual muscle testing (MMT) using the Medical Research Council (MRC) scale for muscle grading shown on next page, although this is not reliable in muscles that are only mildly affected.^{1, 16, 17} Care must be exercised for consistent inter-examiner measures of the antigravity muscles.

Quantitative strength measurements are somewhat more labor intensive, but provide more reliable and reproducible information. These include static (isometric) and dynamic (isokinetic) measurements, most often done in selected muscle groups (usually bilateral knee, elbow, shoulder, and neck flexors and extensors) with a force transducer that displays force output through a digital force monitor. Static pinch strength and grip strength also may be measured using a force transducer, and these measurements followed serially at clinic visits. The highest score from three maximal trials usually is recorded. The handheld dynamometer (HHD) is perhaps the most practical yet reliable way to obtain quantitative strength testing in the clinic. The HHD is a small device equipped with an internal load cell that operates as a force transducer. The HHD is capable of measuring force generated by a subject against the examiner who holds the device firmly against the

MRC Grade Degree of Strength

- 5 Normal strength.
- 5- Barely detectable weakness.
- 4S Same as 4 but stronger than reference muscle.
- 4 Muscle is weak but moves the joint against a combination of gravity and some resistance.
- 4W Same as 4, but weaker than reference muscle.
- 3+ The muscle is capable of transient resistance but collapses abruptly. This degree of weakness is difficult to put into words, but it is a muscle that is able to move the joint against gravity and an additional small amount of resistance. It is not to be used for muscles capable of sustained resistance throughout their whole range of movement.
- 3 Muscle cannot move against resistance but moves the joint fully against gravity. With the exception of knee extensors, the joint must be moved through its full mechanical range against gravity. If a patient has contractures that limit movement of the joint, the mechanical range obviously will be to the point at which the contractures cause a significant resistance to the movement.
- 3- Muscle moves the joint against gravity but not through the full extent of the mechanical range of the joint.
- 2 Muscle moves the joint when gravity is eliminated.
- 1 A flicker of movement is seen or felt in the muscle.
- 0 No movement.

subject and provides stabilization (counter-resistance). The maximum force is recorded. Although it does not replace formal quantitative strength testing, the HHD has reasonably good reliability in NMD for weaker muscle groups and is a good alternative to MMT.

Dynamic strength may be assessed using an isokinetic dynamometer at a fixed speed (e.g., 30 degrees per second) for both concentric (shortening) and eccentric (lengthening) contractions. Flexors and extensors should be evaluated through a full range of motion. Parameters that can be measured include peak torque, total work, work per repetition, peak torque to body weight ratio, joint angle at peak torque, range of motion, and fatigue index (decrement in work performance over the exercise bout).

Joint range of motion (ROM) measurements should be done with a standard goniometer following the protocol used by Brooke and colleagues.⁴ Joints to be evaluated for contractures include elbow and wrist extension, hip adduction for iliotibial band tightness, hip and knee extension, and ankle dorsiflexion. Clinically significant contractures are defined as a reduction in ROM by 20°, except in the antigravity muscles, whose resting position is in full extension (e.g., knee and hip joints). In these joints, even as little as a 7° flexion contracture can result in the center of gravity (COG) falling in an unstable plane (e.g., anterior to the hip joint and posterior to the knee center of rotation). Care must be

taken to measure the ROM of two-joint muscles in position of function (e.g., ankle ROM measured with the knee fully extended).

For many NMD patients spine deformity should be evaluated at every clinic visit. Data obtained at the time of the first patient visit and thereafter should include the presence or absence of spine deformity, any interventions, and the patient's age at time of observation. Radiographs should be reviewed and the results recorded along the guidelines recommended by Carman et al⁵ and Fon et al.¹⁰

Pulmonary function tests (PFT), consisting of forced vital capacity (FVC), forced expiratory volume at 1 second (FEV₁), FEV₁/FVC, maximal voluntary ventilation (MVV), and residual volume (RV) should be done at least yearly and more frequently if clinical indications exist. Maximal inspiratory (MIP) and expiratory (MEP) pressures are helpful and are measured near RV and total lung capacity (TLC), respectively, following the technique described by Black and Hyatt³ using a direct ready dial gauge force meter. Again, three technically satisfactory measurements should be obtained and the maximum reading recorded (Table 2). If clinically indicated, arterial blood gas studies or pulse oximetry should be obtained. Standard 12-lead electrocardiograms (ECGs) should be obtained at 1-year intervals. In some diseases, particularly the myopathies, echocardiograms also may be indicated. Records of respiratory and cardiovascular symptoms and findings should be maintained at each clinic visit. The history should include questions relating to short-

Table 2. FUNCTIONAL TESTING FORM FOR PATIENTS WITH NEUROMUSCULAR DISEASE

- A. Functional grade (arms and shoulders). Select one.
 - 1. Starting with arms at the sides, the patient can abduct the arms in a full circle until they touch above the head.
 - 2. Can raise arms above head only by flexing the elbow (i.e., shortening the circumference of the movement) or using accessory muscles.
- IF 1 OR 2 IS ENTERED ABOVE, how many kg of weight can be placed on a shelf above eye level, using one hand?
 - 3. Cannot raise hands above head but can raise an 8-oz glass of water to mouth (using both hands if necessary).
 - 4. Can raise hands to mouth but cannot raise an 8-oz glass of water to mouth.
 - 5. Cannot raise hand to mouth but can use hands to hold pen or pick up pennies from the table.
 - 6. Cannot raise hands to mouth and has no useful function of hands.
- B. Functional grade (hips and legs). Select one.
 - 1. Walks and climbs stairs without assistance.
 - 2. Walks and climbs stairs with aid of railing.
 - 3. Walks and climbs stairs slowly with aid of railing (over 12 seconds for four stairs).
 - 4. Walks unassisted and rises from chair but cannot climb stairs.
 - 5. Walks unassisted but cannot rise from chair or climb stairs.
 - 6. Walks only with assistance or walks independently with long leg braces.
 - 7. Walks in long leg braces but requires assistance for balance.
 - 8. Stands in long leg braces but unable to walk even with assistance.
 - 9. Is in wheelchair.
 - 10. Is confined to bed.

ness of breath with ambulation, at rest, and during sleep; palpitations; dyspnea; and chest pain. A thorough review should be made of cardiopulmonary complications, including pneumonia, prolonged upper respiratory tract infections, respiratory compromise requiring assisted ventilation, and heart failure.

Functional evaluation levels and timed motor performance (TMP) tests are helpful in determining the extent of disability. Functional classifications utilize the upper extremity scale reported by Brooke et al⁴ and the lower extremity scales used by Vignos et al.^{21, 22} The functional grades, which consist of six levels of function for the upper extremities and ten levels for the lower extremities, are shown in Table 2. Timed motor performance tests should follow the protocol reported by Brooke et al⁴ shown below.

Enter time in seconds.

T = tried but failed to complete by time limit of 120 seconds.

1. Standing from lying supine.
2. Climbing four standard stairs (beginning and ending standing with arms at sides).
3. Running or walking 30 feet (as fast as compatible with safety).
4. Standing from sitting on chair (chair height should allow feet to touch floor).
5. Propelling a wheelchair 30 feet.
6. Putting on a T-shirt (sitting in chair—see instructions).
7. Cutting a 3" × 3" premarked square from a piece of paper with safety scissors (lines do not need to be followed precisely).

Neuropsychologic measurements may be helpful in some of these diseases, particularly if there are educational and vocational problems. However, previous reports that used some of the standard measurement tools suggested that subtle physical impairments may have negatively affected the test results. Therefore, caution is advised in interpreting these tests. Tools such as the Category Test, Seashore Rhythm Test, and Speech-Perception Test should be used, if possible, because performance on these instruments is not dependent on motor function.¹¹⁻¹³

Once the diagnosis is confirmed, the patient and family should be thoroughly educated about the expected outcome and what problems may be encountered. The physiatrist should then assess the patient's and family goals and develop a palliative and rehabilitative program that matches those goals.

Major advances in the understanding of the molecular basis of many NMDs has greatly enhanced diagnostic accuracy and may provide the basis for therapeutic intervention. There have also been major pharmacologic advances in the treatment of some NMDs, particularly ALS.

The physiatrist may become involved in the prescription of disease-altering medications for the various NMDs, and therefore should familiarize him/herself with the appropriate pharmacologic agents available. In addition, if not directly involved in research, the physiatrist should nonetheless encourage enrollment in experimental protocols, which not only furthers science but provides some hope for the patient. Education and employment are very important with respect to self-esteem, quality of life, and integration into the community and should be emphasized in people with slowly progressive NMD. Patients should be referred to a support group. Support groups often are a great resource, not only for psychologic support but for problem-solving and recycling of equipment.

Given the many advances that have occurred in the management of people with NMD, many patients will now live through their childbearing years, possibly having children, and expecting to enjoy a high quality of life. The physiatrist can play a critical role in the comprehensive approach to the management of NMD patients and help them fulfill these expectations.

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CLINICAL APPROACH TO THE DIAGNOSTIC EVALUATION OF PROGRESSIVE NEUROMUSCULAR DISEASES

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Progressive acquired or hereditary neuromuscular diseases are disorders caused by an abnormality of any component of the lower motor neuron—anterior horn cell, peripheral nerve, neuromuscular junction (presynaptic or postsynaptic region), or muscle. The notion that a pathologic abnormality in a neuromuscular disease may be purely isolated to one anatomic region of the lower motor neuron with primary or secondary changes in muscle is true only in selected conditions. Increasingly, we are recognizing neuromuscular diseases that are multisystem disorders. For example, myotonic muscular dystrophy may affect skeletal muscle, smooth muscle, myocardium, brain, and ocular structures; Duchenne muscular dystrophy gives rise to abnormalities of skeletal and cardiac muscle, the cardiac conduction system, and brain; Fukuyama congenital muscular dystrophy affects skeletal muscle and brain; mitochondrial encephalomyelopathies may affect the mitochondria of multiple tissues.

Neuromuscular diseases may be acquired (e.g., amyotrophic lateral sclerosis (ALS), poliomyelitis, Guillain-Barré syndrome, myasthenia gravis, or polymyositis), but the most common etiology is genetic (e.g., spinal muscular atrophy [SMA], hereditary motor and sensory neuropathy [HMSN], congenital myasthenia gravis, or Duchenne muscular dys-

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