

# ***GUIDE TO NEUROMUSCULAR DISEASES FOR REHABILITATION COUNSELORS (revised 08/02)\****

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(revised 08/02)\*

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## ***INTRODUCTION***

In 1990, the estimated number of individuals with major neuromuscular diseases (NMDs) in the United States ranged from 95,000-122,000. Of these individuals, the number of potential candidates for Department of Rehabilitation services ranged from 57,000 - 73,000 (ages 16-64 years, 60%). In 2002, the estimated prevalence is 400,000, a figure that soars to 4.5 million when individuals with post-polio syndrome are included.

Department of Rehabilitation Counselors have unique challenges when working with clients with neuromuscular diseases. Because the NMD population is small compared to other groups with disabling conditions such as heart disease, Rehabilitation Counselors rarely come into contact with individuals with these diseases. Since there are over 200 distinct NMD, it is even more unlikely that a counselor would come into contact with two individuals with the same type of neuromuscular disease. As for the information a client would be expected to provide about their physical condition, many individuals with NMD cannot provide the kind of specific medical details that would allow a Counselor to assist them effectively. A client may not have received a specific diagnosis beyond a disease category such as “motor neuron disease” or “neuropathy,” and the many diseases within each category have different characteristics that influence functional capabilities and quality of life. The rate of progression is highly individualistic and prognoses for work expectancy / life expectancy are often tenuous and contingent upon a complex set of economic, social, and psychological factors.

## ***DIRECTIONS FOR USE OF THIS GUIDE***

The guide has three sections:

- **Section 1:** Descriptions of neuromuscular diseases most likely to be encountered by Rehabilitation Counselors.
- **Section 2:** Disease progression graphs showing progressive levels of physical impairment. This section contains information about:
  - 1) Anterior Horn Cell Diseases: amyotrophic lateral sclerosis; progressive spinal muscular atrophy; progressive bulbar palsy; spinal muscular atrophies.
  - 2) Myoneural Junction Diseases: myasthenia gravis.
  - 3) Peripheral Nerve Diseases: Charcot Marie Tooth syndrome.
  - 4) Muscle Diseases: Becker muscular dystrophy; Duchenne muscular dystrophy; facioscapulohumeral dystrophy; limb girdle syndrome; myotonic muscular dystrophy; polymyositis and dermatomyositis.
- **Section 3:** Vocational implications of neuromuscular diseases at different levels of physical limitation.

The disease descriptions and disease progression graphs are organized by major category and then by specific disease. A glossary of simple genetic terms precedes the disease descriptions.

*Caution:* A counselor's decision on whether or not to provide Department of Rehabilitation services to an individual with a neuromuscular disease is based on many factors. The material is meant to provide information on **some** of those factors.

The disease progression graphs were developed to give Rehabilitation Counselors a "snapshot" of what often happens to individuals with specific neuromuscular diseases. They are designed as a guide for Rehabilitation Counselors rather than as exact medical descriptions. For more complete information, Counselors are advised to visit the nmdinfo web site at <http://www.nmdinfo.net>.

## ***SECTION 1: DISEASE DESCRIPTIONS***

## ***NEUROMUSCULAR DISEASES (NMDs)***

The neuromuscular system includes the anterior horn cells in the spinal cord, the peripheral (motor) nerves, the myoneural junctions (MNJs), and the muscles. Acting as a single functional unit, this system is called the “motor unit.” The anterior horn cells are connected to both the brain and the peripheral nerves. The axons (it may be helpful to picture them as the electric wires that connect telephone poles) in the peripheral nerves are connected to the muscles via the myoneural junctions.

Different neuromuscular diseases can affect one or more components of this unit and may be either acquired or hereditary. The major symptoms are weakness and/or atrophy of skeletal muscle. Examples of NMDs are summarized below.

<b>AFFECTED COMPONENT</b>	<b>ETIOLOGY</b>	
	<b>ACQUIRED</b>	<b>HEREDITARY</b>
<b>Anterior horn cell</b>	Amyotrophic lateral sclerosis	Spinal muscular atrophy
	Poliomyelitis and post-polio syndrome	
<b>Peripheral nerves and motor nerve roots</b>	Physical injury	Charcot Marie Tooth
	Toxins	
	Guillain Barre syndrome	
<b>Myoneural junctions</b>	Myasthenia gravis	Hereditary myasthenia gravis
	Botulism	
<b>Muscle</b>	Polymyositis	Muscular dystrophies
		Congenital and metabolic myopathies

Characteristics common to the major neuromuscular diseases include:

- Weakness and/or fatigue (all NMDs).
- Limb contractures: Duchenne dystrophy, early onset spinal muscular atrophy, Charcot Marie Tooth syndrome.
- Spinal deformity/scoliosis: Duchenne dystrophy, spinal muscular atrophy, Friedreich's ataxia.
- Restrictive lung disease: all NMDs over time (rapidly progressive in Duchenne muscular dystrophy and amyotrophic lateral sclerosis).
- Cardiac dysfunction: Duchenne muscular dystrophy, myotonic muscular dystrophy.
- Cognitive defects: congenital myotonic muscular dystrophy, Duchenne muscular dystrophy (30% of boys with Duchenne muscular dystrophy have some cognitive impairment).

## **DEFINITION OF TERMS IN DISEASE DESCRIPTIONS**

Disease description definitions are taken from the World Health Organization (WHO) classification of disablement. While impairment and disability characterize a person's functional limitations, these definitions don't necessarily define an individual's capabilities or potential. It is often social attitudes and misperceptions that lead to "DISADVANTAGE." The table below shows how these terms are interpreted in cases of neuromuscular disease.

<b>ORGAN</b>	<b>IMPAIRMENT (usually progressive)</b>	<b>DISABILITY</b>	<b>DISADVANTAGE</b>
<b>Skeletal Muscle</b>	Decreased strength and endurance	Decreased motor performance	Decreased quality of life
		Decreased mobility	
		Decreased limb function	
		Increased fatigue	
<b>Bone and Joint</b>	Joint contractures	Decreased function	Decreased educational opportunities
		Increased pain and deformity	
<b>Lungs</b>	Decreased pulmonary function	Increased restrictive lung disease	Decreased employment opportunities
		Increased fatigue	
<b>Heart</b>	Cardiomyopathy	Decreased cardiopulmonary capacity	Increased dependency
	Conduction defects	Increased fatigue	
<b>Central Nervous System</b>	May decrease intellectual capacity		

### **GENETIC GLOSSARY.**

**AUTOSOMAL:** Gene is carried on one of the non-sex chromosomes. In autosomal diseases, both males and females may be affected.

**CHROMOSOMES:** There are 23 pairs of chromosomes in the cells of each individual (46 chromosomes). Of these, females have two X chromosomes and males have one X and one Y (sex chromosomes). NMDs may be inherited as autosomal dominant autosomal recessive, or X-linked (sex-linked) recessive disorders. Genes, the basic units of heredity, are located on the chromosomes.

**DOMINANT:** Abnormal gene masks the normal gene. Dominant diseases usually appear in every generation, and are transmitted by either parent to male or female offspring with a 50% probability of developing the disease.

**RECESSIVE:** Abnormal gene is not expressed unless there is no normal gene to counteract its presence. Recessive diseases tend to skip generations. Autosomal recessive diseases are transmitted to up to 25% of male and female offspring when both parents carry the abnormal gene. Sex-linked recessive diseases are transmitted

through the clinically unaffected female to 50% of the male offspring, who develop the disease, and 50% of the female offspring, who are carriers of the abnormal gene.

### ***DISEASE PROGRESSION***

The neuromuscular disease descriptions (and disease progression graphs) have been organized by major category, and then by specific disease within that category, as follows. (This is not an all-inclusive list of NMDs. It represents those NMDs that would most likely be encountered by Rehabilitation Counselors.) For more complete information about each disease, visit the RehabInfo Network web site at <http://www.rehabinfo.net>.

### ***ANTERIOR HORN CELL DISEASES***

#### **Adult Motor Neuron Diseases.**

##### Type of inheritance:

These diseases are mostly acquired through a sporadic genetic mutation of unknown cause; approximately 10% of ALS cases are the result of family inheritance.

##### Clinical onset:

Typically after 50 years of age.

##### Clinical Description (characteristics):

ALS - anterior horn cells and the central nervous system (CNS) are involved. Onset of weakness and spasticity that is usually preceded by fatigue, cramping and muscle fasciculations (twitching). Usually weakness of pharyngeal muscles with marked difficulty in chewing, swallowing and speaking.

Progressive spinal muscular atrophy (PSMA) - only the anterior horn cells are involved so the only finding is progressive weakness.

PBP (a form of ALS) - manifested by progressive weakness of pharyngeal muscles.

##### Distribution of weakness:

ALS - in most cases weakness is asymmetrical. Typically, the disease begins with painless weakness and wasting of the muscles of one hand or one foot, which becomes generalized and severe. Progressive spasticity accompanies the weakness.

PSMA - progressive weakness.

PBP - progressive weakness of pharyngeal muscles

##### Progression:

ALS - the most common and rapidly progressive anterior horn cell disease. Disease leads to loss of ambulation 1-2 years after onset.

Other forms (above) - progress more slowly.

##### Life expectancy:

ALS - 3-5 years following onset.

Other forms - variable.

##### Treatment:

None.

Medical rehabilitation can improve quality of life.

Training / work outlook:

- ALS - very poor.
- PSMA - with some individuals may be good.
- Others - variable, usually limited.

**Spinal Muscular Atrophies.**

The spinal muscular atrophies (SMAs) consist of a large group of heterogeneous disorders. Some have a proximal distribution of weakness, others distal or generalized. Progression may be rapid to very slow. All are inherited. Most are autosomal recessive, with males usually more severely affected than females.

**Childhood Spinal Muscular Atrophy (SMA Type II)**

Type of inheritance:

Autosomal recessive.

Clinical onset:

Six to 24 months of age.

Distribution of weakness:

Usually generalized but initially may be primarily evident in the proximal muscles of the lower extremities. There is also weakness in arms, legs, upper and lower torso.

Progression:

Usually relatively slow.

Clinical characteristics:

- Delayed motor development landmarks.
- No cognitive defects.
- Loss of ambulation variable.
- 65% incidence of significant contractures.
- 57% have spine deformity.
- Restrictive lung disease common.
- Good psychosocial adjustment.

Life expectancy:

Age at death variable; 70% of individuals are alive at 25 years.

Treatment:

Medical rehabilitation can retard the progression of contractures and spine deformity.

Training / work outlook:

Impairment and disability high. Historical employment rate 63%. Education level 13 +/- 5 years.

**Juvenile Spinal Muscular Atrophy (Kugelberg-Welander syndrome, SMA Type III).**

Type of inheritance:

Usually autosomal recessive.

Clinical onset:

After 18 months of age to early adolescence.

Distribution of weakness:

Symmetrical proximal limb weakness usually commencing in the lower extremities.

Progression:

Slow.

Clinical characteristics:

Loss of ambulation variable.

Low incidence of contractures, spine deformity, restrictive lung disease, and cardiac involvement.

Good psychosocial adjustment.

High incidence of calf hypertrophy.

No cognitive defects.

Life expectancy:

Most have normal life span.

Treatment:

Medical rehabilitation can retard the progress of contractures.

Training / work outlook:

Usually good.

Low impairment, disability, and handicap.

Historical employment rate 63%.

Education level 13 +/- 5 years.

***MYONEURAL JUNCTION DISEASE***

**Myasthenia Gravis (MG).**

Type of inheritance:

An acquired disease.

Two variants: Neonatal Myasthenia.

Congenital Myasthenia.

Clinical onset:

Occurs in all decades of life, most frequently at about age 40.

Among young adults, women more affected than men.

Differential diagnosis:

Main characteristic is muscle fatigue or temporary weakness, rather than permanent loss of strength.

Tendency to recover after rest.

Most common presenting symptoms relate to weakness of eye muscles, causing ptosis or diplopia.

Distribution of weakness:

Fatigue (temporary weakness) is generalized

Progression:

At onset, may last only a few days, then disappear, only to return weeks or months later.

If after 1-2 years, myasthenia is still restricted to the ocular muscles, it is unlikely to become generalized.

Clinical characteristics:

Difficulty chewing.

Dysarthria and dysphagia common.

Since there is primary fatigue rather than weakness, muscle atrophy does not occur except in patients with a severe chronic course.

The nature of the disease in any given patient is usually established within a few months of onset, with fluctuations afterward.

Life expectancy:

Mortality from Myasthenia is less than 10%.

Treatment:

Good control of disease and symptoms with proper medical treatment.

Training / work outlook:

Impairment, disability and handicap low, with proper treatment.

Work outlook is good for most individuals.

## *PERIPHERAL NERVE DISEASES*

### **Hereditary Motor and Sensory Neuropathy (HMSN).**

**Charcot Marie Tooth syndrome (CMT) is the most common type of HMSN.**

Type of inheritance:

Most are autosomal dominant.

Clinical onset:

Usually first and second decade of life.

Distribution of weakness:

Distal weakness usually starting in the foot dorsiflexors (foot drop) and progressing to the hand and forearm muscles and foot plantar flexors.

Rarely involves upper arm, shoulder, thigh or hip muscles until late in the disease.

Progression:

Usually slow.

Clinical characteristics:

Primary distal distribution of weakness.

Associated sensory loss, usually mild and late in the disease.

Most patients can ambulate, with short leg braces, long after the onset

16% (low) incidence of contractures, mostly pes cavus.

29% spinal deformity, mostly kyphosis.

Restrictive lung disease in some cases.

Cardiac involvement rare.

No cognitive defects.

Good psychosocial adjustment.

Life expectancy:

Most have normal life span.

Treatment:

Medical rehabilitation can prevent or retard the progress of weakness and contractures.

Training / work outlook:

Work outlook is good.

Low impairment, disability and handicap.

Historical employment rate 67%.

Educational level 12 +/- 3 years.

## ***OTHER PERIPHERAL NERVE DISEASES***

### **Hereditary Motor Neuropathy (HMN) (with subtypes)**

### **Hereditary Sensory and Autonomic Neuropathy (HSAN) (with subtypes)**

#### Type of inheritance:

Most autosomal dominant.

#### Characteristics:

Most inherited types of peripheral nerve diseases have a primarily distal distribution of weakness.

#### Progression:

Variable, but usually slow.

## ***MUSCLE DISEASES***

### **Becker Muscular Dystrophy (BMD).**

#### Type of inheritance:

Sex (X)-linked recessive.

#### Clinical onset:

Typically 6-20 years of age.

#### Differential diagnosis:

Precise methods for diagnosis and carrier detection available.

Can be confused with the rare late onset Duchenne muscular dystrophy, limb girdle syndrome with early onset, and SMA Type 3.

#### Distribution of weakness:

Similar to Duchenne muscular dystrophy (DMD).

#### Clinical characteristics:

Cognitive impairment not a characteristic.

Similar to Duchenne muscular dystrophy with delayed course. 43% with mild contractures.

Spine deformity rare.

Restrictive lung disease rare.

Cardiac involvement common.

Good psychosocial adjustment.

#### Rate of progression:

Usually slow.

Inability to ambulate in most cases at 15-25 years after onset.

#### Life expectancy:

Often normal life span.

#### Treatment:

Medical rehabilitation valuable for reducing complications.

#### Training / Work outlook:

Usually good work outlook.

Impairment, disability, handicap low.

Historical employment rate 80%.

Educational level 12 years +/- 2.

## **Duchenne Muscular Dystrophy (DMD).**

### Type of inheritance:

Sex (X)-linked recessive.  
Transmitted through unaffected females to males.  
50% probability male offspring will be affected.  
50% probability female offspring will be carriers.

### Clinical onset:

Age range 1-5 years.

### Differential diagnosis:

Precise methods for diagnosis and carrier detection are now available.  
DMD can be confused with Becker's dystrophy, and limb girdle syndrome of early onset with calf hypertrophy.

### Distribution of weakness:

Early selective involvement of hip muscles rapidly followed by weakness of the shoulder girdle muscles.

### Clinical characteristics:

Cognitive impairment in 30% of cases.  
Other early development normal followed by lordosis (sway back); waddling gait; toe walking; difficulty rising; difficulty climbing stairs.  
Involvement usually symmetrical. Ultimately involves all skeletal muscles.  
Calf hypertrophy until nonambulatory.  
74% have severe progressive contractures.  
63% have scoliosis.  
81% have significantly reduced pulmonary function, restrictive lung disease and cardiac involvement.

### Rate of progression:

Consistently progressive.  
Loss of ambulation age 9-11 years.  
Death usually from respiratory complications (18-25 years old).

### Treatment:

Medical rehabilitation can improve quality of life.

### Training / Work outlook:

Extremely limited.  
Impairment, disability, handicap high.  
Usually poor DR candidates.

## **Fascioscapulohumeral Muscular Dystrophy (FSHD; Landouzy-Dejerine)**

### Type of inheritance:

Autosomal dominant.  
Transmitted by either parent to children of both sexes with 50% probability of incidence.

### Clinical onset:

Range 7-25 years of age.

### Differential diagnosis:

Sometimes confused, early in disease, with the ocular myopathies, myasthenia gravis (MG), congenital ptosis, and congenital facial diplegia.

Distribution of weakness:

Initial involvement of face muscles (lack of facial mobility).  
Initial involvement of shoulder girdle muscles (difficulty raising arms overhead).  
Leads to forward slope of shoulders.  
Subsequent slow spread to hip girdle muscles.  
Distal arm / hand muscles affected late in disease.  
Often asymmetric weakness and atrophy.

Progression:

Usually very slow progression of weakness.  
No apparent progression for several years, in some cases.  
Loss of ambulation uncommon.

Clinical characteristics:

Considerable variation among and within families.  
Unlined face, pouting appearance of lips, difficulty closing eyes and inability to whistle.  
Occasionally have indistinct speech.  
33% incidence of mild contractures.  
Significant restrictive lung disease in some cases.  
35% with spine deformity (usually hyperlordosis).  
No calf hypertrophy.  
Cognitive impairment not associated with this disease.  
Good psychosocial adjustment.

Life expectancy:

Normal.

Treatment:

May benefit from medical rehabilitation.

Training / Work outlook:

Impairment, disability and handicap low.  
Historical employment rate 88%,  
Educational level 14 years +/-3.

**Limb Girdle Muscular Dystrophy (LGMD; limb girdle syndrome).**

Type of inheritance:

Autosomal recessive is most common. About 10% of cases representing at least six subtypes are autosomal dominant.

Clinical onset:

Generally before age 20 for autosomal recessive forms.

Differential diagnosis:

Prior to availability of genetic testing, many disorders presenting with muscle weakness in a limb-girdle distribution led to diagnostic confusion, including spinal muscular atrophy (SMA) and the mitochondrial and metabolic myopathies. LGMD has also been mistaken for fascioscapulohumeral muscular dystrophy and congenital muscular dystrophy.

Distribution of weakness:

Patients with LGMD generally show weakness and wasting restricted to the limb musculature, proximal greater than distal. Most patients with LGMD show relative sparing of the heart and bulbar muscles, although subtypes 1B (laminopathy) and 2C-2F (sarcoglycanopathies) have an associated cardiomyopathy that may be the predominant feature, rather than muscle weakness.

Progression:

LGMD can present as a severe muscle disease beginning in infancy, or it may manifest and progress slowly during adulthood. Data now suggest that severity is related to the type of mutation. Wheelchair dependency at 5<sup>th</sup> decade or later is common.

Clinical Characteristics:

Spine deformity, restrictive lung disease and cardiac involvement are relatively rare.  
No facial involvement or cognitive impairment.  
Good psychosocial adjustment all subtypes.

Life expectancy:

Usually normal with good medical management of secondary complications.

Treatment:

Medical rehabilitation can improve quality of life.

Training / work outlook:

Impairment, disability and handicap low in adult-onset subtypes.  
Work outlook can be poor with LGMD onset in childhood.  
Educational level 13 +/- 2 years.  
Historical employment rate 71%.

**Myotonic Muscular Dystrophy (Steinert's disease)**

Type of inheritance:

Autosomal dominant.  
Transmitted by either parent to children of both sexes with 50% probability of incidence.

Clinical onset:

Weakness at age 5-35 years.  
Myotonia usually occurs before weakness.

Differential diagnosis:

Sometimes confused with congenital myotonia and paramyotonia during early stages of the disease.

Distribution of weakness:

Weakness and atrophy usually generalized but primarily proximal in early stages with later weakening of the hand and forearm muscles.  
Facial muscles are sometimes involved.

Progression:

Variable but usually very gradual

Clinical characteristics:

Myotonia (inability to relax muscles) and stiffness primarily noted in hand muscles.  
Multiple organ system involvement  
19% with significant contractures.  
15% with deformity of the spine.  
Restrictive lung disease in some cases.  
High incidence of cardiac involvement, especially life threatening conduction defects.  
Often have indistinct speech.  
May have cataracts.

Life expectancy:

Variable, depending on severity of cardiac involvement.

Treatment:

None for weakness.

Myotonia and heart conduction defects may be reduced by drug treatment.

Cataracts can be corrected with surgery.

Training / work outlook:

Impairment and disability usually low, but handicap high, apparently due to poor psychosocial adjustment.

Usually poor work outlook

Historical employment rate 39%

Educational level 13 +/- 2 years.

## **Polymyositis and Dermatomyositis Syndromes**

Type of inheritance:

Acquired disease that is more common in females.

Five types:

Adult polymyositis.

Dermatomyositis.

Childhood polymyositis.

Myositis with malignancy.

Myositis associated with other connective tissue diseases.

Clinical onset:

At any age, but usually second to fifth decade.

Differential diagnosis:

May be confused in early stages with limb girdle muscular dystrophy.

Distribution of weakness:

Primarily severe (acute) and proximal.

Weakness of anterior neck and pharyngeal muscles very common.

Usually symmetrical

Progression:

Most types are rapidly progressive without treatment. With the exception of the childhood type and myositis with malignancy, most patients respond well to drug therapy.

Clinical characteristics:

Weakness present in all cases.

Over 50% experience pain or muscle tenderness.

33% arthritic features.

Contractures and muscle atrophy rare (except in cases of long-standing resistance to treatment).

Occasional cardiac interstitial fibrosis and pneumonitis in acute stage.

Disease usually self-limiting with successful treatment.

Life expectancy:

May be fatal in childhood polymyositis. Normal with successful treatment.

Treatment:

Drug therapy usually successful

Training / work outlook:

Impairment, disability and handicap high only in chronic form.

Good work outlook in most cases but poor in chronic form and childhood type.

***SECTION II: DISEASE PROGRESSION GRAPHS.***

## *Definition of Terms*

### *Disease Progression Graphs*

The following definitions are guidelines only, since the diseases can progress in an erratic manner. Note: A Department of Rehabilitation client receiving SSI Disability payments or SSDI benefits is classified as "severely disabled" by Department of Rehabilitation / Rehabilitation Counselors regardless of the level of physical restriction.

#### A. Mild.

1. Limited debilitating effects.
2. Does not limit, or limits to a minor degree:
  - a. Standing.
  - b. Walking.
  - c. Use of hands and arms.
  - d. Fine finger dexterity.
  - e. Full time employment.
  - f. Exerting 50 to 100 pounds of physical force (if otherwise large and old enough).

#### B. Moderate.

1. Physical functioning is reduced to a level where a new occupation needs to be developed or considered.
2. The individual is able to function in medium (occasionally), light or sedentary occupations.
3. Most individuals travel and function independently.
4. Usually good candidates for DR.

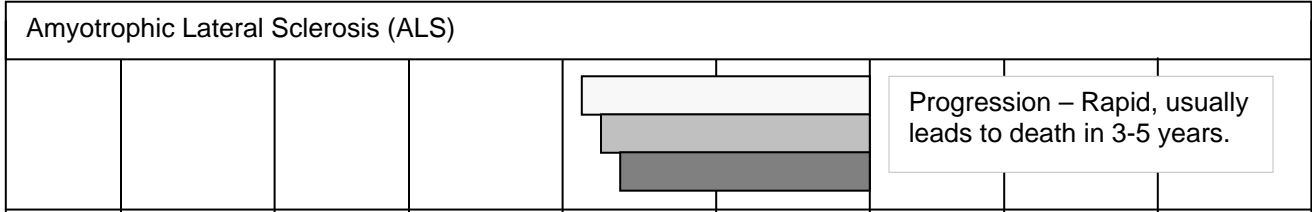
#### C. Substantial (Severe).

1. Individuals with neuromuscular diseases are automatically classified "severely disabled" by Department of Rehabilitation, according to federal regulations.
2. When a physician indicates the limitations are substantial or very physically limiting.
3. Unable to use public transportation (paratransit exception).
4. Unable to perform sustained work activity for six hours or more.
5. Have disfigurement or deformity so pronounced as to cause social rejection.
6. Have speech unintelligible to non-family members.
7. Unable to climb one flight of stairs or walk 100 level yards without pause.
8. Has so little manual dexterity or coordination as to be unable to button buttons or write intelligibly.

<p>Graph bars indicate ages at which an individual may experience a particular level of physical involvement. The three bars taken together represent the possible variations in disease progression for one individual. Beginning and end ages on each bar are approximate and vary from individual to individual. Beginning (left end) of "mild" bar is the earliest onset of the disease. The right end of the "substantial" bar is the work expectancy limit (maximum age 65.)</p>	<p>Levels of physical involvement:</p> <p>Mild <span style="display: inline-block; width: 20px; height: 15px; background-color: #e0e0e0; border: 1px solid black; margin-left: 10px;"></span></p> <p>Moderate <span style="display: inline-block; width: 20px; height: 15px; background-color: #a0a0a0; border: 1px solid black; margin-left: 10px;"></span></p> <p>Substantial <span style="display: inline-block; width: 20px; height: 15px; background-color: #606060; border: 1px solid black; margin-left: 10px;"></span></p>
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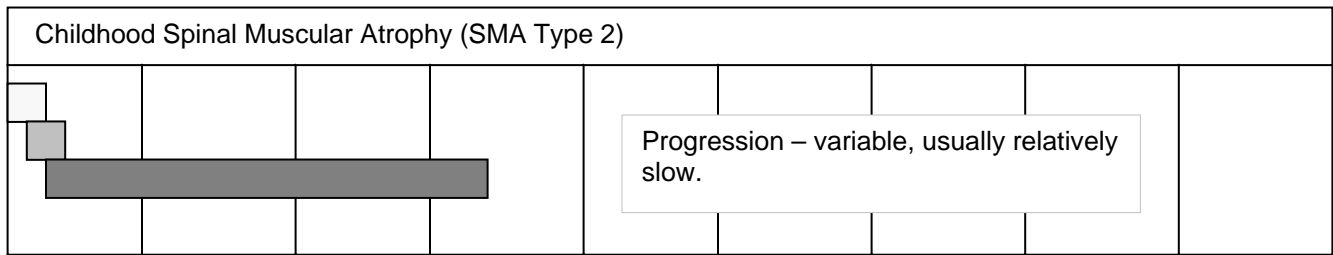
Age in Years

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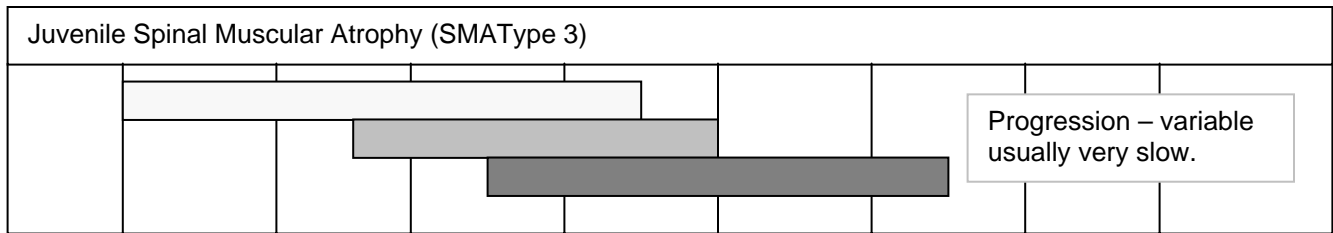
Age In Years

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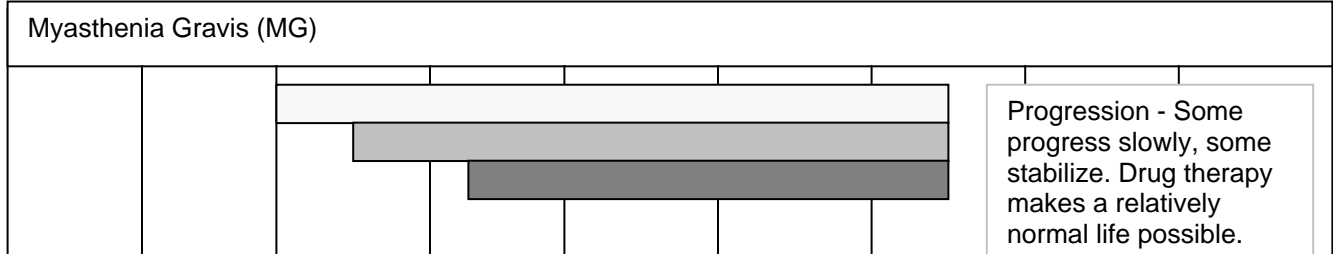
Age In Years

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Age In Years

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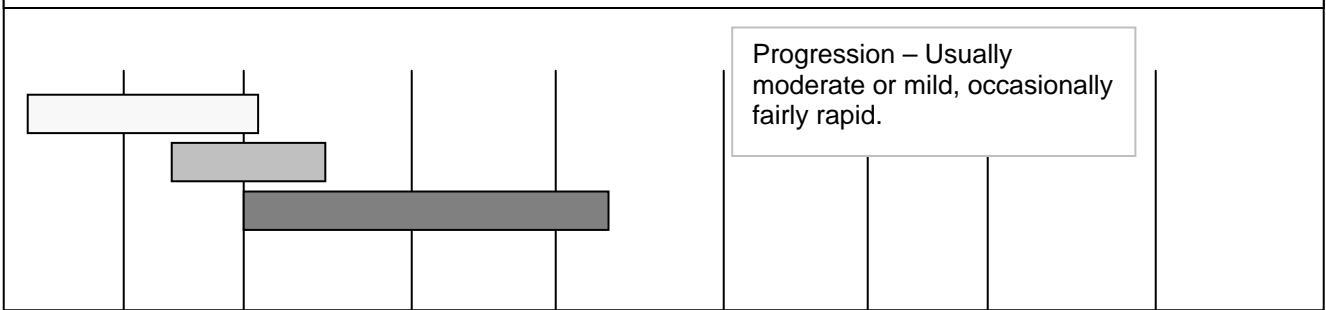
Hereditary Motor & Sensory neuropathy (HMSN) (Charcot Marie Tooth, CMT)



Age In Years

0 10 20 30 40 50 60 70 80 90

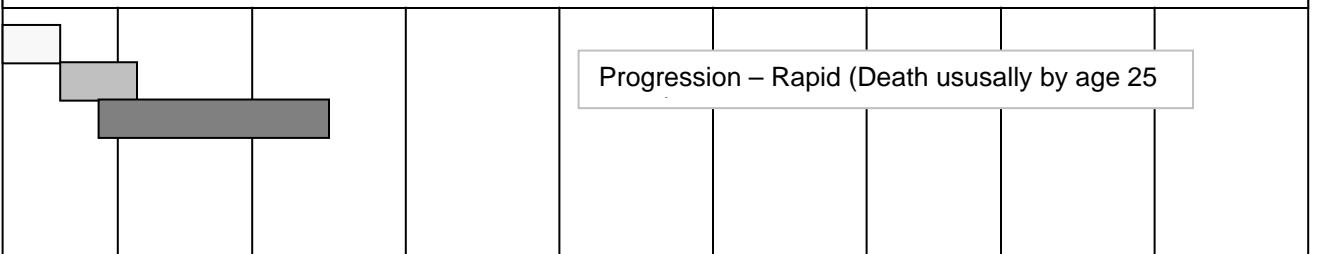
Becker's Muscular Dystrophy (BMD)



Age In Years

0 10 20 30 40 50 60 70 80 90

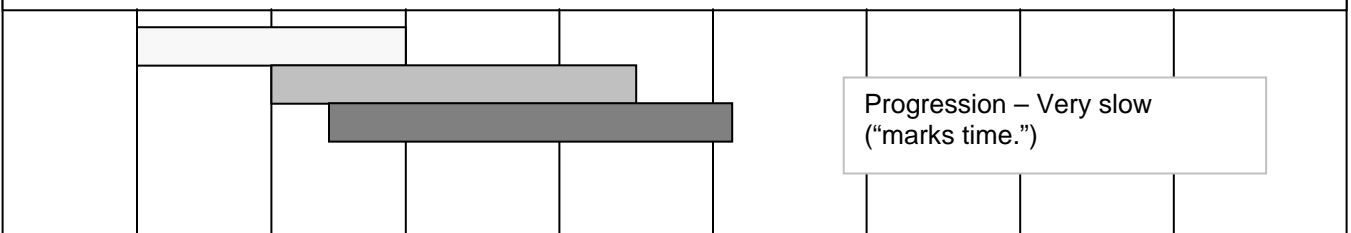
Duchenne Muscular Dystrophy (BMD)



Age In Years

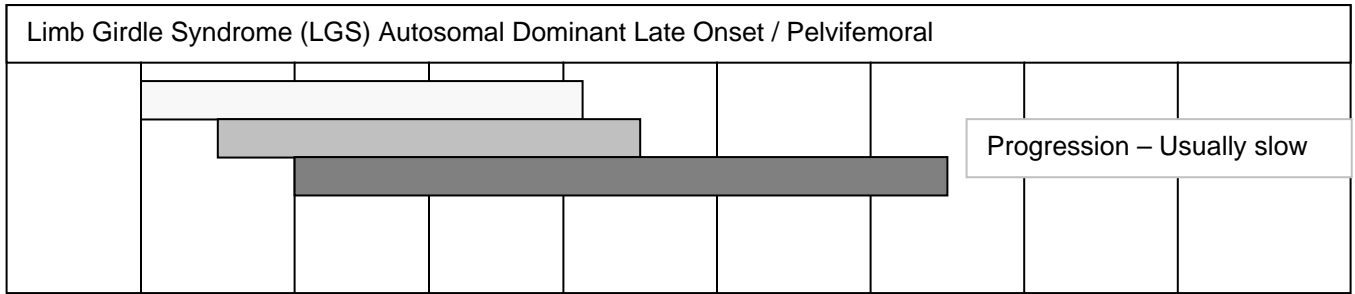
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Facioscapulohumeral Muscular Dystrophy (FSH)



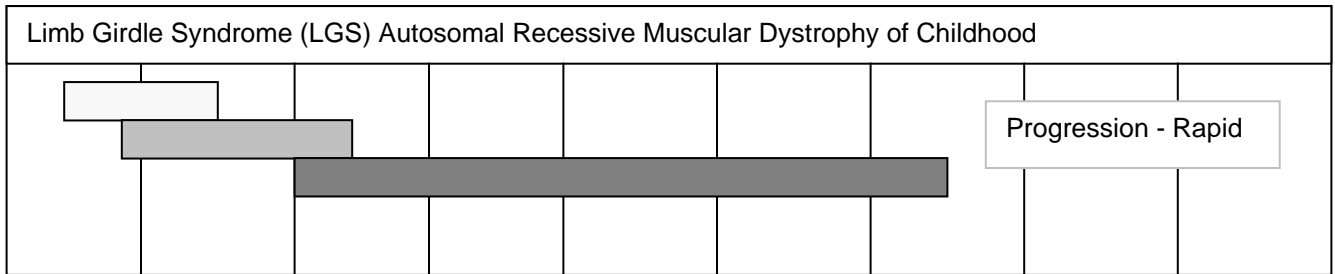
Age in Years

0 10 20 30 40 50 60 70 80 90



Age In Years

0 10 20 30 40 50 60 70 80 90

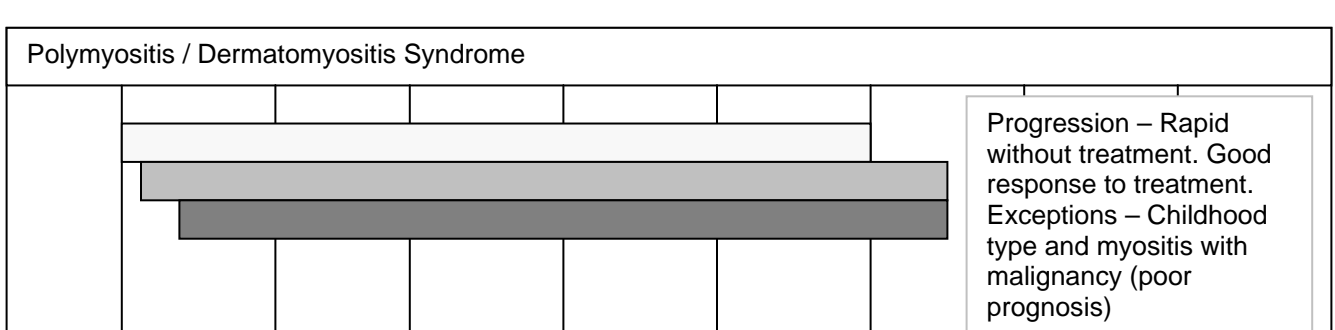


0 10 20 30 40 50 60 70 80 90



Age In Years

0 10 20 30 40 50 60 70 80 90



***SECTION 3: VOCATIONAL IMPLICATIONS.***

## *Vocational Implications*

The use of the Dictionary of Occupational Titles (DOT) requires some understanding of the total Job classification processes. These are as follows:

### Summary Listings of Occupational Categories, Divisions, and Groups

#### Occupational Divisions:

- (000 through 199) Professional, Technical or Managerial Occupations
- (200 through 299) Clerical and Sales Occupations
- (300 through 399) Service Occupations
- (400 through 499) Agricultural, Fishery, Forestry, and Related Occupations
- (500 through 599) Processing Occupations
- (600 through 699) Machine Trades Occupations
- (700 through 799) Benchwork Occupations
- (800 through 899) Structural Work Occupations
- (900 through 999) Miscellaneous Occupations

#### Term Titles and Definitions:

These are listed in DOT, Fourth Edition, 1977 (pp. 5-14). These were used to select appropriate jobs for individuals with NMDs.

#### Occupational Group Arrangement:

These are listed in DOT, Fourth Editions, 1977 (pp. 15-946). Definitions in this section were used to select appropriate jobs for individuals with NMDs.

#### Glossary.

Listed in DOT, Fourth Edition, 1977, (pp. 947-963).

#### Alphabetical Index of Occupational Titles:

Listed in DOT, Fourth Edition, 1977 (pp. 965-1156).

#### Occupational Titles Arranged by Industry Designation:

Listed in DOT, Fourth Edition, 1977 (pp. 1157-1367).

#### Physical Demands:

See Selected Characteristics of Occupations Defined in the Dictionary of Occupational Titles, 198 1, Appendix A, (pp.465-466.)

Used to classify jobs for individuals with NMDs.

#### Environmental Conditions:

Listed in Selected Characteristics of Occupations Defined in the Dictionary of Occupational Titles, 198 1, Appendix B, (pp. 467).

#### Specific Vocational Preparations-Training Time:

Listed in DOT Supplement, Appendix D, 1981 (pp. 473).

Physical Demands:

Listed in DOT Supplement to Fourth Edition, Appendix D, 1986 (pp. 101-102).

Environmental Conditions:

Listed in DOT Fourth Edition Supplement, Appendix D, 1986 (pp. 103).

The job listings for NMDs (at the end of this guide) have been abstracted from the DOT and include ratings for "physical demands " and "environmental conditions."

The ratings are.

1. S. - Sedentary Work
2. L. - Light Work
3. M. - Medium Work
4. H. - Heavy Work
5. V. - Very Heavy Work
6. For environmental conditions see Appendix B, D.O.T., 1981, p.467.

These ratings are helpful when selecting occupations for the general public. For individuals with NMDs it was necessary to break down the light and medium occupations into subcategories which are consistent with physical / mental limitations imposed by NMD.

### ***VOCATIONAL PLACEMENT***

Individuals with NMDs are placed in work ability categories, and these are matched with jobs and job areas. They are:

1. Unable to work, or unable to work for 2 years or more: If an individual cannot work for 2 years or more, they are not considered a rehabilitation candidate. The rehabilitation process often takes 1-2 years to complete. Disabilities in this category include, but are not limited to:

Amyotrophic lateral sclerosis (ALS).

Duchenne's muscular dystrophy (DMD).

Polymyositis (if prognosis is for rapid and fatal progression).

2. Very limited employment potential - little to no use of extremities (occasionally can use voice, head or mouth/teeth to perform job activity). Must have attendant and/or multiple assistive devices to function. Severely limiting cognitive impairment. Rapidly progressing syndrome (work potential 2-4 years). Usually high risk for rehabilitation failure. Disabilities in this category include, but are not limited to:

Limb girdle syndrome (LGMD) (when syndrome has early onset - 1st decade in life - or rapid progression)

Becker's (BMD) muscular dystrophy (with early onset).

Myotonic muscular dystrophy (MMD) (if condition has early onset and/or is rapidly progressive).

Polymyositis (when severe and chronic).

Charcot Marie Tooth syndrome (with early onset--5-15 years and rapid progression)

### 3. Sedentary work.

Juvenile spinal muscular atrophy (in later stages).

Hereditary motor and sensory neuropathy (Charcot-Marie-Tooth disease) (in later stages at later age)

Facioscapulohumeral (FSH) (in later stages at later age).

Limb Girdle Syndrome (LGS) (progression of disease variable, usually in later stages at later age)

Becker's muscular dystrophy (BMD) (between working age 16 and debilitated age, usually mid 20s)

Myotonic Muscular Dystrophy (MMD) (in later stages usually age 35+)

Polymyositis (when chronic and severe)

Dermatomyositis (same as Polymyositis)

Spinal Muscular Atrophy of adults (SMA) (usually after age 45)

Myasthenia Gravis (in later stages, often 30s in women, 50s in men)

### 4. Light work.

Facioscapulohumeral (FSH) (in middle stages of disease, can usually work at this level for many years).

Limb girdle muscular dystrophy (same as above)

Becker's muscular dystrophy (BMD) (can occasionally function at this level for a few years - ages 16-24 usually).

Myotonic muscular dystrophy (MMD) (in middle stages of the disease, usually before age 35).

Polymyositis (mild and chronic).

Dermatomyositis (same as polymyositis above).

Juvenile spinal muscular atrophy (in middle stages, can usually function at this level for many years).

Spinal muscular atrophy of adults (SMA type 4) (can function at this level during middle stages of the disease).

Charcot Marie Tooth disease (CMT), Hereditary motor sensory neuropathy (at this work level middle stages of disease, for few to many years).

Myasthenia gravis - (at this work level as long as fatigue factor remains low).

5. Medium work.

Limb girdle syndrome (when progression remains slow and disability remains slight).

Muscular dystrophy of late onset (MD) (fits this category when onset is late and disability is slight) polymyositis (with mild / late onset).

Dermatomyositis (same as polymyositis above).

Spinal muscular atrophy (can work at this level in early stages).

Charcot Marie Tooth disease, hereditary motor sensory neuropathy (in early stages with slow progression)

6. Heavy work: None

7. Very heavy work: None

***COMMENTARY: NEUROMUSCULAR DISEASES AND JOB PLACEMENT***

Social attitudes and preconceptions play an important role in how people with disabilities are perceived. In the past, misinformation and stereotypes have governed how individuals with neuromuscular disease are treated across a broad set of situations. This has begun to change but there is still a long way to go. The foregoing was intended to provide some insight into the functional capacity of those who live with neuromuscular disease and enable you to better serve this segment of the community.

The mission of the Department of Rehabilitation (DR) is to “assist Californians with disabilities in obtaining and retaining employment and maximizing their ability to live independently in their communities.” However, the ability of the DR to serve the NMD population has not been entirely successful. Among individuals with NMD who have been referred to the DR in the past, only 18% wanted to be referred again and of those who had not been referred only 30% were interested in a referral.<sup>1</sup> This implies that people who have experience with the DR were not entirely satisfied, and those with no experience did not appear to believe DR services would be beneficial. So, how can rehabilitation counselors better serve this segment of the community?

Unemployed disabled individuals who want to and believe that they are able to work cite low-paying jobs and the lack of education, training and skills needed for full-time work as a major cause of their unemployment. This does not however mean that they cannot successfully complete training programs or, with some adaptations in the work environment, succeed in high level positions. Through effective functional assessment, education, and the use of assistive technology, the functional consequences of NMD on career success can be greatly reduced and in some cases even eliminated.

## ***IMPROVING ATTITUDES AND UNDERSTANDING***

Improved service begins with an accurate understanding of what a person with neuromuscular disease can do and what their limitations are. Currently, societal attitudes and lack of understanding are among the most significant barriers individuals with NMD face. Perhaps the best way to convey the impact that stereotypes and misinformation have on persons with NMD is to hear it in their own words:

A participant in one RRTC study remarked, "Physical barriers did not turn out to be the problem.... Social attitudes were the most profound barriers throughout the course of my life." (interview #4, 3/17/95).

Another reported, "My biggest barriers have been my own attitudes to begin with. And then when I worked on that attitude and became very healthy with where I was as a disabled person, it became even more glaring, the attitude problems that society has with disability." (interview #36, 8/12/96).

It is important to understand that the needs of people with a slowly progressive NMD change over time, which requires a periodic reassessment by the rehabilitation team of assistive technologies that can improve mobility and speech.

Perceived limitations can play an important role in how a person with NMD is placed in an employment setting. By educating themselves on the basic characteristics of NMD or even just spending a little extra time with each person discussing specifically how the disease affects them, counselors can develop a better understanding of the barriers a person with NMD faces. More important is to figure out how to work around those barriers for a successful career placement outcome. Remember that the most common barriers a person with NMD faces are related to lack of mobility, not cognitive deficits.

### ***COUNSELOR RESPONSIBILITIES AND ASSISTIVE TECHNOLOGY (AT)***

Successful career placement requires the expertise and cooperation of everyone involved in the rehabilitation process, including the employee, their doctor, employer, and rehabilitation technology specialist, but it must begin with an accurate assessment by the rehabilitation counselor. An elementary understanding of NMD would contribute significantly to a counselor's ability to assess an individual's competitiveness in the job market. Also, focus on what people are able to do and what they could be capable of doing if the proper training and assistive technology were available to them.

Assistive technology was defined in the Technology Related Assistance for Individuals with Disabilities Act of 1988 as:

"any item, piece of equipment, or product system, whether acquired commercially off the shelf, modified or customized, that is used to increase, maintain, or improve functional capabilities of individuals with disabilities."

Rehabilitation professionals must develop a general awareness of the potential for assistive technology for individuals with NMD. Comprehensive knowledge of AT is helpful but not required. However, the counselor must know enough to recognize when AT is appropriate and provide the necessary resources

or referrals. Selection, implementation, and follow up all contribute significantly to the utility of adaptive technology and the difference it makes in a person's ability to succeed in a career. Assistive technology can greatly increase independence, productivity, and confidence, all of which contribute to an individual's success in the employment marketplace.

The most effective application occurs when a multidisciplinary team works together to develop solutions to resolve barriers to employment. This team should include the rehabilitation counselor, the client, an expert in assistive devices, and the employer. Having someone on the team who knows the potential sources of funding for AT can contribute to a client's success in obtaining AT. However, solutions do not need to be expensive. A close partnership between the rehabilitation counselor, employer, and employee can create cost-effective solutions that benefit both the employee and employer.

The rehabilitation counselor is also in a good position to facilitate and coordinate effective communication between the members of the rehabilitation team. A counselor can address potential concerns of employers and dispel any misconceptions they may have regarding accommodation of a disabled individual. For example, some employers assume that adaptations are expensive and time consuming, when most accommodations are very simple and have a minimal cost.

Agencies must look for innovative ways to provide AT expertise. This can be accomplished several ways. Someone who has a strong technical background in assistive technology can be hired as part of the regular staff. Counselors can attend training programs or attend assistive technology conferences to improve their understanding of how to effectively integrate assistive technology into the rehabilitation process. Another option is to use a program like Project Threshold.

Project Threshold at Los Amigos Research and Education Institute, Inc. at Rancho Los Amigos National Rehabilitation Center is one example of how a range of services can be provided efficiently to clients who are experiencing problems as they pursue independent living, education or vocational goals. This program provides services to clients throughout the state of California. Occupational therapy and engineering staff provide evaluations, recommendations, custom modification of equipment and follow-up. These services are available in most areas, but what makes Project Threshold unique is that they come to the individual's home and office. This in turn provides an accurate assessment, because they can identify barriers specific to an individual's home and work environment and allows hands-on testing of technology before recommendations are made.

Obtaining feedback is a critical component of successfully completing the job placement process. The current low satisfaction rate with the DR might be improved by revising the way in which clients give feedback in order to elicit more constructive suggestions about the job placement process.

Another tool that may contribute to successful job placement is the new Occupational Information Network (O\*NET). This is a database that contains comprehensive information on job requirements. O\*NET replaces the Dictionary of Occupational Titles and offers a more dynamic, interactive framework for exploring career options. By identifying and describing the key components of occupations, including the high-tech industry, O\*NET supplies up-dated information critical to the effective training, education, counseling and employment of workers.<sup>2</sup>

People with neuromuscular disease have shown a remarkable ability to adapt to their disease. Integrating new resources, like O\*Net, with assistive technologies, combined with an increased level of awareness on the part of the rehabilitation counselor, could contribute significantly to long term, fulfilling careers for individuals with neuromuscular disease. However, an individual with NMD must also make a significant contribution to the career placement process.

### ***CONSUMER RESPONSIBILITIES***

The consumer has much to do with the success of the job placement process. The consumer must provide detailed information about what their needs are and be prepared to advocate on their own behalf. Each person going through the rehabilitation process must make a strong effort to educate him or herself, because without a basic awareness of the resources and services that are available, the ability to overcome barriers to independence, productivity, and professional success will be limited. Consumers must also be willing to provide constructive feedback about the effectiveness of AT and other services. Without this feedback, improvements to existing services and technology will be limited. By playing an active role in the rehabilitation and job placement process, an individual with NMD will increase the chance that they will find a fulfilling and lasting career.

### ***CONCLUSION***

Currently, societal attitudes and lack of understanding are among the most significant barriers individuals with neuromuscular disease face. In the past, misinformation and stereotypes have governed how individuals with neuromuscular disease are treated, especially in the employment marketplace. We hope that, by increasing awareness about NMD, we have dispelled some of these stereotypes and are contributing to a change in attitude that will improve the quality of life and job opportunities for individuals with neuromuscular disease.

### **REFERENCES.**

<sup>1</sup> Employment Profiles in Neuromuscular Diseases, American Journal of Physical Medicine & Rehabilitation. Am J Phys Med Rehabil 76: 26-37, 1997.

<sup>2</sup> O\*Net, <http://online.onetcenter.org/>