RCS 6080
Medical and Psychosocial Aspects of Rehabilitation Counseling

Neuromuscular Disorders

Multiple Sclerosis

- MS is characterized by exacerbations and remissions of a multitude of signs and symptoms indicative of damage to several areas of the brain and spinal cord
- MS is a nonhereditary chronic disease of the CNS, with onset mostly in young adult life
  - In most cases, symptoms begin between the ages of 20 and 40, although onset before age 10 and after age 60 have been reported
  - There seems to be a genetic predisposition to the disease that is modified by some environmental influence

Multiple Sclerosis

- MS is rare in some parts of the world and more common in others – it increases in frequency with latitude in both northern and southern directions
  - Worldwide, MS occurs with much greater frequency in higher latitudes (above 40° latitude) away from the equator, than in lower latitudes, closer to the equator.
  - In the U.S., MS occurs more frequently in the northern states than in southern states. Nationwide, there are an estimated 400,000 people with MS.

Multiple Sclerosis

- An individual who is born in an area with a higher risk of developing MS and moves to an area of lower risk, acquires a risk similar to that of the new home if the move occurs prior to adolescence.
- MS is more common among Caucasians (particularly those of northern European ancestry) than other ethnic groups, and is almost unheard of in some populations, such as Inuit.
- MS is 2-3 times as common in women than in men.

Multiple Sclerosis

- Certain outbreaks or "clusters" of MS have been identified, but the cause and significance of these outbreaks is not known.
- In certain populations, a genetic marker, or trait, has been found to occur more frequently in people with MS than in those who do not have the disease. Thus far, no specific gene has been identified that definitely confers susceptibility to MS. Large-scale research is ongoing to identify the multiple genes that appear to make people susceptible to MS.

Multiple Sclerosis

- The exact cause of MS is unknown, but the most widely held theory is that MS occurs in people who have a genetically determined increased immune response to viral infections and that some part of the immune response "attacks" the myelin sheath covering the different components of the CNS
  - The resulting pathological picture is that of scattered areas of demyelination (plaques) in the brain and spinal cord
  - These plaques can often be seen with an MRI scan

Multiple Sclerosis

- There are four major areas of research focus:
  - Immunology: new mechanisms are being tested for their ability to influence immune function, particularly the function of T cells, which clearly play a role in MS
  - Genetics: Although MS is not directly inherited, it seems that a person must carry a genetic predisposition if he or she is to develop MS. Investigators are screening the genetic makeup of families in which more than one member has MS. This may someday help identify the genes inherited by people who are susceptible to MS.

Multiple Sclerosis

  - Virology: MS is believed to be triggered by something in the environment. Although many scientists now suspect that no single virus causes MS, the key may lie in the way a person genetically predisposed to MS handles viral infections.
  - The Biology of the Glial Cells: Myelin is manufactured by the glial cells of the CNS. Understanding how these cells function, how they form myelin, and how they might form new myelin after disease (which is the best hope for recovery of function) is an important and growing area of MS research.

Multiple Sclerosis

- The clinical course of MS is chronic, lasting for decades
  - The onset of exacerbations is acute, and remission can occur within days
  - The first exacerbation is almost always followed by complete recovery, and subsequent attacks are gradually less completely resolved
  - With each subsequent attack, there may be a recurrence of old symptoms and some additional ones

Multiple Sclerosis

- The signs and symptoms vary in nature and severity, depending on the area of injury in the CNS and the chronicity of the illness
  - The most common are fatigue, muscle weakness and spasticity, impaired sensation and coordination, unexplained pain, visual disturbances, gait abnormalities, bowel and bladder dysfunction, mental status changes, dizziness and vertigo, and emotional problems
  - A typical characteristic of MS is that all of the symptoms tend to vary in both nature and severity with time

Multiple Sclerosis

- MS tends to take one of four clinical courses, each of which might be mild, moderate, or severe
  - Relapsing-Remitting MS (RRMS): a relapsing-remitting course characterized by partial or total recovery after exacerbations
    - This is the most common form of MS
    - 70-75% of people with MS initially begin with a relapsing-remitting course
  - Secondary-Progressive MS (SPMS): a relapsing-remitting course, which later becomes steadily progressive
    - Attacks and partial recoveries may continue to occur
    - Of the 70-75% who start with RRMS, more than 50% will develop SPMS within 10 years and 90% within 25 years

Multiple Sclerosis

  - Primary-Progressive MS (PPMS): a progressive course from onset
    - The symptoms generally do not remit
    - 15% of people with MS are diagnosed with PPMS, although the diagnosis usually needs to be made after the fact when the person has been living with MS for a period of time with progressive disability but not acute attacks
  - Progressive-Relapsing MS (PRMS): a progressive course from the onset, which is also characterized by obvious acute exacerbations
    - It is quite rare – 6-10% of people with MS appear to have PRMS at diagnosis

Functional Disability of MS

- All aspects of function can be affected by disability, including ambulation, transfers, ADLs, vision, hearing, and mental status
  - Gait difficulties are the most common and can include spasticity, ataxia, loss of position sense, and weakness
  - Arm and hand function can be similarly affected, and there is often an intention tremor, which further makes self-care activities difficult or impossible
  - Bladder functions are affected
  - Deteriorating vision, hearing, and speech, along with mental depression or unrealistic euphoria, can limit a person's social interaction

Treatment and Prognosis of MS

- Medical management of MS is two-fold
  - One is the use of medications to arrest exacerbations and possibly delay or moderate recurrent symptoms
  - The second is the maintenance of function and prevention of physical and functional deterioration, using a multidisciplinary rehab team approach
- From the textbook – the average survival of people with disabilities is 35 years
- From the National MS Society –most people with MS can expect 95% of the normal life expectancy
- Death is usually secondary to respiratory, renal, or decubitus ulcer infections

Vocational Implications of MS

- People who fall into the RRMS and PPMS categories can be expected to work for 25 years past onset or longer
- Modifications to the work environment may be needed as they begin to use orthotics, assistive devices for ambulation, hearing aids, eyeglasses, upper extremity splints, and other devices
- People with jobs requiring heightened physical activity, such as walking, prolonged standing, and exertion of physical force, would benefit from vocational retraining early in the disease course

Vocational Implications of MS

- Although a person may be expected to lose time during exacerbations, the time lost may not be excessive
- Disease stability may be more important than disease severity
  - The person's functional status 5 years after diagnosis is probably the best indicator of future performance
  - People who have relapses and remissions are reported to have higher employment rates than those with a progressive course

Vocational Implications of MS

- Some research findings indicate that the following reasons were given for a person's unemployment:
  - Spastic paresis, incoordination, bowel and bladder dysfunction (Bauer, Firnhaber, & Winkler, 1968)
  - Physical difficulty, visual difficulty, transportation difficulty, fatigue (Scheinberg et al., 1981)
  - Mobility (LaRocca et al., 1982)

Vocational Implications of MS

- Several specific precautions should be taken for almost all people with MS who return to work
  - In general, the most important precaution concerns the temperature of the environment in which the person works
  - Workers with MS should follow a program of energy conservation and should take frequent rest breaks during activities
    - Use of assistive devices
    - Do most demanding work during most energetic hours

Muscular Dystrophy

- The muscular dystrophies are a group of progressive hereditary diseases characterized by muscle weakness, muscle loss, joint contractures, and deformity
- The three main types of MD are Duchenne, facioscapulohumeral (FSH), and myotonic

Duchenne MD

- X-linked hereditary disorder occurring in males only
- The prevalence rate is 1.9-3.4 per 100,000 worldwide
- Symptoms first appear in early childhood, before age 3, and are characterized by a waddling gait and difficulties in climbing and running
  - The calves and upper arm muscles are overdeveloped, a condition known as pseudohypertrophy
  - As muscle weakness and muscle wasting progress, a characteristic posture of toe walking, with bent knees and an increased lumbar lordosis, is assumed
  - A progressive scoliosis of the thoracic and lumbar spine occurs in many cases

Duchenne MD

- Mild to moderate mental retardation is common
- If the ability to ambulate is maintained beyond age 12, the condition is Becker's MD, a more slowly progressive form of the disease
- The clinical course is rapidly progressive - most people with Duchenne MD are unable to walk or care for themselves as a result of severe muscle weakness by early adolescence
  - Death from respiratory failure usually occurs toward the end of the second decade

FSH MD

- A group of syndromes inherited in an autosomal dominant fashion and differing from each other in extent of clinical expression
- Prevalence is 0.2-0.5 per 100,000
- Onset of symptoms occurs during adolescence and includes upper arm and shoulder girdle weakness, impaired eye and lip closure, and eventual footdrop
- There is no intellectual deficit
- Near-normal life expectancy because of the chronic, slowly progressive disease course, and ambulation is often preserved

Myotonic MD

- An autosomal dominant hereditary disorder with varied clinical expression, affecting males more than females
- Prevalence is 5 per 100,000
- Symptoms include an inability of the muscles to relax after a forced contraction (myotonia); loss of muscle power and bulk in the face and neck, leading to poor head control; a flat facial expression; swallowing difficulties; and weakness of the distal extremities
  - Eyes are affected by cataracts
  - There are cardiac arrhythmias and gastrointestinal motility problems

Myotonic MD

- Mild mental retardation and personality disorders have been noted
- Onset of symptoms is in the early adult years (20-30), with facial, hand, and foot weakness appearing first
- Myotonic MD is slowly progressive
  - People with mild muscle involvement may not become disabled and will have a normal life expectancy
  - If muscle weakness is severe, the person will become incapacitated in the fourth or fifth decade of life

Functional Disability and MD

- In all three forms, muscle weakness and muscle wasting lead to impaired ambulation, arm function, and general mobility

Treatment and Prognosis of MD

- There is no specific drug treatment for the muscular dystrophies
- PT is essential early on in Duchenne MD to prevent muscle contractures and maintain muscle power
- OT is essential to provide assistance with ADLs, home equipment, and wheelchair fitting, including specialized seating arrangements, and the accommodation of a portable ventilator unit
- In myotonic MD, speech therapy can offer maintenance of the muscles of mastication and deglutition, as well as alternative modes of communication when speech is no longer possible

Vocational Implications of MD

- Vocational retraining is appropriate in Becker's MD, keeping in mind the chronic yet slow progression toward muscle weakness
- People with FSH MD and myotonic MD may be able to sustain their chosen vocations or, if more severely affected, may need job retraining that accommodates future upper-body and upper-extremity weakness
- Social skills training is useful as an integral part of a comprehensive, specialized educational program if the person with MD is socially immature

Vocational Implications of MD

- Weakness decreases an individual's capacity to perform a job that requires heavy manual labor
- Changes in a person's mobility also have a direct effect on job performance
- The rehabilitation counselor must assess the potential for overwork weakness syndromes during the evaluation and planning stages of a vocational program
- Visual defects and other complicating medical problems can able affect job placement and performance

Amytrophic Lateral Sclerosis

- Amyotrophic lateral sclerosis (ALS) is a chronic disease of middle to late adult life, affecting the voluntary motor pathways of the CNS
- Symptoms usually begin after age 40

Amytrophic Lateral Sclerosis

- It is characterized by muscle weakness and fasciculations (involuntary contraction or twitching of muscle fibers)
  - The clinical course is rapidly progressive, beginning with muscle atrophy and loss of power and fasciculations in the extremities and face and progressing to muscle spasticity and severe weakness
  - Symptoms include gait abnormalities, arm function deficits, impaired speech and swallowing mechanisms, and respiratory muscle weakness
  - Intellectual functions are not affected

Amytrophic Lateral Sclerosis

- There are four widely accepted subgroups of ALS
  - Sporadic
    - This type may affect anyone, anywhere affecting all genders and races
    - It has a prevalence rate of 4-6 per 100,000 people
    - It is the most common form of ALS in the United States - 90 to 95% of all cases
  - Familial
    - Occurring more than once in a family lineage (genetic dominant inheritance)
    - In these families, there is a 50% chance each offspring will inherit the gene mutation and may develop the disease
    - Accounts for a very small number of cases in the United States - 5 to 10% of all cases

Amytrophic Lateral Sclerosis

- Guamanian
  - An extremely high incidence of ALS was observed in Guam and the Trust Territories of the Pacific in the 1950's - 50 to 100 times greater
  - One theory as to the possible reason is the exposure to a toxin from the cycad nut
- Secondary
  - ALS-like symptoms have been associated with syphilis, hypoglycemia, and plasma cell disorders

Functional Disability and ALS

- Gait difficulties, loss of arm muscle power, and fatigue are common early complaints, requiring a variety of assistive devices, including lower extremity braces and upper extremity splints to maintain posture and assist in function
- As symptoms progress, trunk and neck braces and specialized feeding devices are necessary

Functional Disability and ALS

- The person rapidly becomes dependent on a caretaker, progressing to complete loss of functional independence
- Loss of speech function, combined with the inability to write and eventually to the inability to use computerized communication aids, leads to physical and mental isolation in spite of intact intellect and mental status

Treatment and Prognosis of ALS

- In spite of trials with different agents, there is no specific drug treatment for ALS
- Spasticity of extremity muscles and those involved in chewing and swallowing is reduced with medication
- PT and OT are necessary from the onset to aid in ADLs and to maintain muscle range and power
- Speech therapy for improved food intake and swallowing, as well as training in alternative modes of communication, is essential early on

Treatment and Prognosis of ALS

- From textbook: Death usually occurs within 5 years of onset, from respiratory failure, aspiration of oral contents, or infection
- From ALS website:
  - The life expectancy of a person with ALS averages about two to five years from the time of diagnosis
  - 50% of all affected live more than 3 years after dx
  - About 20% of people with ALS live 5 years or more and up to 10% will survive more than 10 years and 5% will live 20 years
  - There are people in whom ALS has stopped progressing and a small number of people in whom the symptoms of ALS reversed

Vocational Implications of ALS

- For a younger person diagnosed with ALS, a vocational counselor ideally should intervene with the employer to keep the person on the job while accommodating his or her special needs, such as rest periods and the increased use of computerized tools
- Heavy physical labor could not be sustained by a person with ALS, therefore retraining should be done

Charcot-Marie-Tooth Syndrome

- Charcot-Marie-Tooth syndrome is a slowly progressive hereditary disease of the peripheral nervous system
- It is mostly transmitted in an autosomal dominant fashion and occasionally is autosomal recessive or X-linked
- It is characterized by muscle weakness and deformity of the feet and hands

Charcot-Marie-Tooth Syndrome

- Symptoms are usually noted late in the first decade or early in the second decade of life
- The prevalence is 2-5 per 100,000
- The disease is caused by a peripheral nervous system defect - a loss of the myelin sheath of the peripheral nerves with damage to the exposed nerves and replacement of all by scarlike tissue

Charcot-Marie-Tooth Syndrome

- Symptoms
  - Congenital foot deformities may be the only symptoms in some families
  - In others, loss of leg muscle bulk, footdrop, and a "stocking and Glove" loss of sensation occurs early
  - Hand deformity caused by a loss of the small muscles occurs later on
  - Some people may have spinal deformities and a tremor
  - Cognitive and mental functions are not affected

Functional Disability and CMT

- Congenital foot deformities may be mild and not interfere with the development of ambulation
- The more severe deformities and those progressing to bilateral foot weakness create difficulty in walking
- Eventual loss of hand muscle power will cause difficulties with daily living and work activities and necessitate functional retraining

Treatment and Prognosis of CMT

- There is no known drug treatment
- PT is necessary as soon as orthotics and assistive devices are introduced
  - The goal is to retain ambulation and preserve joint range of motion, as well as to protect the sound joints from damage and deformity
- OT is important in preventing hand and arm deformity and in retaining function
- Life expectancy is normal
- Most people remain ambulatory until old age

Vocational Implications of CMT

- Because onset generally occurs before the age of vocational training, the rehabilitation counselor should be instrumental in planning for a vocation that does not require extensive ambulation or generalized physical exertion and can be carried out in spite of eventual hand weakness and deformities

Parkinson's Disease

- Parkinson's disease is the major cause of neurological disability in people over 60 years of age
- It is a nonhereditary chronic disease of the brain, characterized by abnormal movement and posture
- Symptoms usually begin between the ages of 50 and 65, although rare cases of childhood onset are known

Parkinson's Disease

- It affects both genders and all races equally
- Prevalence throughout the world is 100-150 per 100,000
- The underlying brain dysfunction responsible for the disease symptoms is loss of dopamine and the destruction of the substantia nigra
- The clinical course is progressive, leading to a steady decline in function after the first 3 years

Parkinson's Disease

- The characteristics symptoms include a tremor described as "pill rolling," muscle rigidity described as "cog wheeling," motor slowness, and a tendency to be suddenly "frozen" in one position
  - There are changes in body posture affecting the trunk, hands, and feet
  - The gait is characterized by small, rapid, shuffling steps known as a festinating gait
  - Functions controlled by the autonomic nervous system are affected, resulting in poor temperature control, episodes of hypotension and syncope, and inadequate bowel and bladder emptying
  - Psychological disturbances range from cognitive, perceptual, and memory deficits to frank dementia

Functional Disability and Parkinson's Disease

- In 70% of cases, tremors is the initial complaint
- It is often preceded by a decrease in facial and eye movements and a tendency to remain in one postural position for a long time
- With the onset of muscle rigidity and slowness, the person rapidly deteriorates to a state of total dependency on a caretaker
- All ADLs may require 10 times the normal duration

Functional Disability and Parkinson's Disease

- Walking is so slow and laborious, interrupted by periods of "freezing" in place, that an assistive device and close supervision are necessary
- Voice volume and speech production are affected, and with the earlier loss of writing ability, communication is severely challenged
- Drooling and swallowing difficulties affect eating and lead to weight loss and further debility
- The eventual deterioration of intellectual function, affecting memory and cognition, and in some cases leading to dementia, requires confinement to the home under constant supervision

Treatment and Prognosis of Parkinson's Disease

- The medical treatment consists of lifelong administration of certain medications, rehabilitation, and psychotherapeutic support
- The rehabilitation team is involved in increasing mobility via use of assistive devices, prescribing supportive home equipment, providing training in self-care activities, preventing and managing joint flexion contractions and decubitus ulcers, and prolonged communication

Treatment and Prognosis of Parkinson's Disease

- Prior to the use of medication, 25% of people with Parkinson's died within 5 years, and 80% died within 15 years
  - Levodopa has reduced the mortality rate by 50% and has increased survival by several years

Vocational Implications of Parkinson's Disease

- People will be able to continue working in most vocations that are not extremely demanding physically
- Occupations that involve heavy manual labor or shifts in posture should be changed if onset is in the early 50s and the person is not near retirement

Friedreich's Ataxia

- Friedreich's ataxia is a common, rapidly progressive hereditary disease of the brain and spinal cord
- It is characterized by loss of coordination in the voluntary muscles of the extremities, trunk, and speech apparatus
- Symptoms begin during the first decade or early second decade of life (between 7 and 13), but may be present in infancy
- The disease is passed from parent to offspring in an autosomal recessive fashion

Friedreich's Ataxia

- It occurs worldwide, in all races, and is more common in males
- The prevalence in Europe and North America is 1-2 per 100,000
- The course of Friedreich's ataxia is rapidly progressive, except in a few cases in which early symptoms are arrested and no new ones develop

Friedreich's Ataxia

- The first symptom to occur is usually loss of coordination in the legs, known as gait ataxia
- This is followed by hand or body tremors, known as titubations, and a speech disturbance described as "scanning speech"
- Leg muscle weakness leading to paralysis and muscle atrophy occur later
- Loss of position sense and other sensations in the leg and trunk make it difficult for the person to stand, walk, and sit
- Cardiac abnormalities are common

Friedreich's Ataxia

- The rate of seizures is higher than that in the general population
- In most cases, intelligence is normal to high, but MR and dementia have been noted in some people
- Almost 75% of people with Friedreich's ataxia are born with clubfoot, and 80% are born with scoliosis and kyphosis of the spine
- In most cases, complete loss of independent function occurs 10 to 15 years after onset
- Death may be sudden, secondary to cardiac complication, or may result from an infection following complete physical deterioration

Functional Disability and Friedreich's Ataxia

- Walking is usually affected from the start
- Advanced ataxia, paralysis, and loss of speech necessitate total dependence on a caretaker

Treatment and Friedreich's Ataxia

- There is no known drug for the treatment of Friedreich's ataxia
- During the early phase and in the chronic, less rapidly progressive cases, PT is indicated for maintenance of muscle range and strength

Vocational Implications and Friedreich's Ataxia

- In people with less progressive courses of the disease, vocational and educational guidance is indicated
- The goal is to help the person choose a vocation that is intellectually appropriate yet not demanding in terms of physical strength and coordination

Additional Resources and Information from the Web

- National Multiple Sclerosis Society (www.nmss.org)
- North Florida Chapter of the National MS Society (www.msnorthfl.org)
- JAN – Accommodating People with MS (www.jan.wvu.edu/media/MS.html)
- Muscular Dystrophy Association (www.mdausa.org)
- JAN – Accommodating People with MD (www.jan.wvu.edu/media/MD.html)

Additional Resources and Information from the Web

- ALS Society (www.alsa.org)
- MDA - information on ALS (www.mdausa.org/disease/als.html)
- JAN – Accommodation Ideas for People with ALS (www.jan.wvu.edu/soar/other/als.html)
- Charcot-Marie-Tooth Association (www.charcot-marie-tooth.org/site/content/)
- MDA - information on CMT (www.mdausa.org/disease/cmt.html)
- JAN – Accommodation Ideas for People with CMT (www.jan.wvu.edu/soar/other/cmt.html)

Additional Resources and Information from the Web

- The National Parkinson Foundation, Inc. (www.parkinson.org)
- The Parkinson Association of Southwest Florida, Inc.(in Naples, FL) (www.pasfi.org)
- Parkinson's Disease, Movement Disorders and Rehabilitation Center (in Port Charlotte, FL) (www.parkinson.org/bonsecours.htm)
- JAN – Accommodating People with Parkinson's (www.jan.wvu.edu/media/PD.html)
- MDA - information on Friedreich's Ataxia (www.mdausa.org/disease/fa.html)