

# Neuromuscular Disorders

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Includes Muscular Dystrophy

## *Description of the Disability*

Neuromuscular Disorders is a catchall term for a large number of diseases involving muscle weakness or muscle wasting. They can involve problems with the nerves of the spinal cord or peripheral nervous system, or problems with the muscle fibers themselves. However, they all lead to a gradual weakness and breakdown of the muscles, and to a common set of physical symptoms. In addition to weakness, these common symptoms can include physical (sometimes painful) changes in the joints, difficulty with speech, and difficulty with mobility and fine motor skills.

Many people with neuromuscular disorders face significant issues with **contracture**. Because they have difficulty moving parts of their body, the muscles in those parts are not stretched as often and can begin to grow shorter. This can limit the range of motion of joints and, in some cases, make the person unable to straighten some joints from a bent position.

Each of the many neuromuscular conditions has a distinct name, root cause, severity of symptoms, etc. Physicians sub-classify the neuromuscular conditions into six categories: Muscular Dystrophies, Inflammatory Myopathies (also called myositis), Motor Neuron Diseases, Neuromuscular Junction Diseases, Peripheral Nerve Diseases, and Other Myopathies.

- **Muscular Dystrophies (MD)** - These are usually genetic, although researchers have found the specific genes for only some of the different types. As the MD progresses, fat and connective tissues may replace the person's muscle fibers. This makes the person seem to have full, strong muscles, even though the muscles are actually shrinking dramatically.
  - **Duchene (Pseudohypertrophic MD)** - The most common childhood form of MD, it involves a gradual generalized weakness and wasting of all voluntary muscles.
  - **Myotonic** (also called **Steinert's MD**) - The most common adult form, which starts with an inability to relax the muscles of the hand, feet, neck and face, but slowly progresses to the rest of the body.
  - **Becker MD** - Similar to Duchenne but less severe. However, it can affect the heart.
  - **Congenital MD** - as the name implies, it is present from birth and involves a lack of muscle tone. There can be joint deformities and decreased mental functions.
  - **Distal MD** (also called Miyoshi) - A late midlife onset MD, with a slow progression through the hands, forearms and lower legs. It is not life threatening.
  - **Emery-Dreifuss MD** - Involves the upper arms, shoulders, and calves. It may affect the heart rhythm and require a pacemaker.
  - **Facioscapulo-humeral MD** - Involves the face, shoulders, and upper arms. The person may have trouble opening their eyes or moving their mouth and lips.

- **Limb-Girdle MD** - Involves the shoulders and the pelvis, as well as heart and breathing problems. In advanced stages, the person may need a wheelchair.
- **Oculo-pharyngeal MD** - Primarily involves the face (especially the eyelids) and difficulty swallowing. Onset in late middle life.
- **Inflammatory Myopathies** (Also known as **Myositis**) - These are autoimmune diseases affecting voluntary muscles (See entry on Autoimmune Disorders). Inflammation weakens the muscles and limits the person's ability to stand or walk. These disorders often co-exist with other autoimmune disorders.
  - **Dermatomyositis (DM)** - It is named for a distinctive bluish-purple rash on the upper body and knees, but the significant symptoms are weakness in the neck and limb muscles, and pain and swelling of muscles throughout the body. There is usually a rapid onset of symptoms.
  - **Inclusion Body Myositis (IBM)** - This disorder affects all voluntary muscles, and specifically causes problems with swallowing and gripping. Onset is late and progression is slow.
  - **Polymyositis (PM)** - Affects muscles in the center of the body, including the pelvis and shoulders, but it can involve the person's limbs. Sometimes the person has difficulty swallowing. Adult onset.
- **Motor Neuron Diseases** - These diseases affect the motor nerves in the spinal cord. Some types are inherited, but not all. Initial symptoms include difficulty swallowing, limb weakness, difficulty walking, difficulty speaking, and general muscle weakness. In advanced stages, the person may have trouble breathing.
  - **Amyotrophic Lateral Sclerosis (ALS, also called Lou Gehrig's Disease)** - causes muscle twitches and cramps, progressive muscle wasting, and eventual paralysis.
  - **Spinal Bulbar Muscular Atrophy (SBMA) (also called Kennedy's Disease, X-linked SBMA)** - involves weakness in the tongue, leading to problems with speech and swallowing.
  - **Spinal Muscular Atrophies** - a group of diseases affecting the voluntary muscles, including those of breathing.
- **Neuromuscular Junction Diseases** - These diseases involve problems transmitting signals from the nerve to the muscle.
  - **Congenital Myasthenia Gravis** - a genetic condition with early onset.
  - **Lambert-Eaton Syndrome (LES)** - an autoimmune disease affecting the hips and thighs.
  - **Myasthenia Gravis (MG)** - an autoimmune disease affecting voluntary muscles. Symptoms include double vision, droopy eyelids, and problems chewing, swallowing, talking, and breathing. Activity makes the symptoms worse, rest makes them better.
- **Peripheral Nerve Diseases** - These diseases primarily affect the extremities.

- **Charcot-Marie-Tooth Disease (CMT)** (also called **Peroneal Muscular Atrophy**) - Usually involves the person's feet, lower legs, hands, and forearms. There may also be reduced vision or hearing and scoliosis.
- **Dejerine-Sottas Disease (DS)** - This inherited condition delays a person's motor skills and can cause sensory and mobility problems.
- **Friedreich's Ataxia (FA)** - Rare genetic disease that causes shaky movements and unsteadiness due to impaired limb coordination with weakness and muscle wasting. Speech, coordination and heart problems may be associated.
- **Other Myopathies** - disorders that don't fit the categories above.
  - **Central Core Disease (CCD)**- A rare inherited disorder involving poor muscle development and possible skeletal deformities.
  - **Mitochondrial Myopathies** - These disorders involve problems with the batteries of the body cells, mitochondria. In addition to muscle weakness, there can be heart problems, dementia, seizures, and sensory disorders.
  - **Myotonia Congenita (MC) and Paramyotonia (PP)** - These disorders involve muscles that do not relax quickly after contraction. The person may also have sensitivity to cold.
  - **Periodic Paralysis** (Hypokalemic and Hyperkalemic) - These disorders involve disruption of the calcium biochemistry of the nerves, causing temporary paralysis that can last from 15 minutes to days.

### ***Incidence Statistics***

- More than 50,000 people in the United States have one of the forms of Muscular Dystrophy
- There are approximately 20,000 people living with ALS
- There are between 20,000-36,000 people living with Myasthenia Gravis.
- 100,000-125,000 people in the United States are living with Charcot-Marie-Tooth Disease

### ***Common Treatments, Medications, and Side Effects***

Because there are so many different types of neuromuscular disorders, there are many different types of treatment. In general, the treatments focus on relieving the symptoms of a person's specific disorder. People with contracture problems may use physical therapy, orthotics, and, in some cases, surgery. People with heart problems may need pace makers or heart medications (see entry on Cardiovascular Disease). Some people take Phenytoin, Dantrolene, or Quinine to help with muscles that will not relax quickly after contraction (see drug entry on Antispasmodics / Antimyotonics for side effects). People with some of the autoimmune neuromuscular conditions may take immunosuppressant drugs (see Drug entry for side effects).

### ***Possible Functional Issues***

- Reduced coordination; clumsiness; frequent falling

- Difficulty with fine motor skills
- Limited mobility
- Difficulty talking
- Difficulty swallowing
- Difficulties Breathing
- Fatigue/weakness

## ***Initial Interview Considerations***

### Initial Questions

- What specific tasks cause difficulty for the person?
- Besides fatigue and muscle weakness, what other symptoms does the person experience?
- What times of day are better or worse for the person?
- What triggers, if any, have they notice that make their symptoms flare up?
- What, if anything, helps reduce their symptoms? (rest, exercise, staying warm, etc.)
- What accommodations has the person tried? Which were particularly helpful?
- How long, if at all, can the person stand or walk?
- How difficult is it for the person to write?
- How difficult is it for the person to use a computer? How well can they use voice recognition software to operate a computer?
- Does physical activity relieve the person's symptoms or make them worse?
- How easily do they get tired in general? Would working all day be a challenge?
- Does the person have any sensitivity to cold?
- If the person has associated heart problems, what restrictions, if any, has their doctor put on their activities?
- What is a typical weekend like for the person? (gets at other interests, skills)
- How does the person feel about meeting the public? Would they rather work with a small group of people or a large group?
- How well, if at all, can the person drive a car?
- How stable is their condition? What do they expect their symptoms to be in 5 years?

### Initial Observations

- Does the person use a wheelchair or any mobility aids?
- Does the individual have difficulty grasping writing implements to fill out paperwork?

- How legible is the person's handwriting?
- How easy is it to understand the person's speech?
- Does the person seem to become tired easily?

#### Interview Accommodations (if any)

- Provide a relatively high chair for the person to sit in. Low chairs are harder to get up from.
- If the person has difficulty writing, offer to provide a written summary of the meeting.
- Make sure the room is a comfortable temperature in case the person is sensitive to cold.

### ***Possible Accommodations and Assistive Technology***

- Wheelchair, scooter, or walking aid
- Adjustable work station
- Automatic doors
- Desk or workstation near facilities the person will use often (copier, mail room, bathrooms, etc.)
- If person is greeting the public at a counter, provide a stool to sit on.
- Low pile carpet for easier walking.
- Changes in floor level indicated by a visual and texture change
- Arm rests for computer stations
- Personal attendant at work to help with grooming, eating, etc.
- A designated note taker for meetings
- Adaptive phones, including features like voice activation, large buttons, automatic dialing, and headsets
- Alternative access features for computers, including speech recognition, trackballs, key guards, alternative keyboards, mouth sticks, etc.
- Doors with levers/easy grasp
- Gripping handles on tools
- Flexible schedule to help with fatigue, public transportation issues, etc.
- Frequent breaks
- Home-based employment
- Communication aids
- Alternative communication such as e-mail, faxes, instant messaging, paging, etc.

- A space heater or extra blanket if person is sensitive to cold

### ***Career Planning Issues***

- For some neuromuscular disorders, exercise helps reduce symptoms, but for others exercise makes symptoms worse. It may be possible to structure the work setting or schedule to match this aspect of the person's condition.
- Some people with a neuromuscular disorder experience sensitivity to heat and cold, especially cold.
- Some neuromuscular disorders progress quickly and some progress slowly. It is important to consider the rate the person is experiencing and prepare supports for the future.
- The symptoms of neuromuscular disorders are often very visible to others. This may make some people with neuromuscular disorders uncomfortable around the stares of the general public. They may prefer job settings that only include a small group of people with whom they can become comfortable.

### ***Emerging Issues***

- Causes, treatments

### ***Additional Information Resources***

- Muscular Dystrophy Association - [www.mdausa.org](http://www.mdausa.org)
- Muscular Dystrophy Family Foundation, Inc.- [www.mdff.org](http://www.mdff.org)
- National Institute of Neurological Disorder & Stroke - [www.ninds.nih.gov](http://www.ninds.nih.gov)
- Trover Foundation- [www.stayinginshape.com](http://www.stayinginshape.com) — health and exercise information specifically focusing on neuromuscular diseases.
- Job Accommodation Network - [www.jan.wvu.edu](http://www.jan.wvu.edu)