
Neuromuscular disorders

Summary

- The combination of the nervous system and muscles, working together to permit movement, is known as the neuromuscular system.
 - There are many diseases that are classified as neuromuscular disorders.
 - Some of the known causes include inherited disorders, hormonal disorders and autoimmune disorders.
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The brain controls the movements of skeletal (voluntary) muscles via specialised nerves. The combination of the nervous system and muscles, working together to permit movement, is known as the neuromuscular system.

If you want to move part of your body, a message is sent to particular neurons (nerve cells), called upper motor neurons. Upper motor neurons have long tails (axons) that go into and through the brain, and into the spinal cord, where they connect with lower motor neurons. At the spinal cord, the lower motor neurons in the spinal cord send their axons via nerves in the arms and legs directly to the muscle they control.

A typical muscle is serviced by anywhere between 50 and 200 (or more) lower motor neurons. Each lower motor neuron is subdivided into many tiny branches. The tip of each branch is called a presynaptic terminal. This connection between the tip of the nerve and the muscle is also called the neuromuscular junction.

The electrical signal from the brain travels down the nerves and prompts the release of the chemical acetylcholine from the presynaptic terminals. This chemical is picked up by special sensors (receptors) in the muscle tissue. If enough receptors are stimulated by acetylcholine, your muscles will contract.

There are many diseases that are classified as neuromuscular disorders.

Symptoms of neuromuscular disorders

The symptoms of neuromuscular disease vary according to the condition and may be mild, moderate or life threatening. Some of these symptoms may include:

- Muscular weakness
- Muscle wastage
- Muscular cramps
- Muscle spasticity (stiffness), which later causes joint or skeletal deformities
- Muscle pain
- Breathing difficulties
- Swallowing difficulties.

Causes of neuromuscular disorders

Some of the causes may include:

- Genetic mutation
- Viral infection
- Autoimmune disorder
- Hormonal disorder
- Metabolic disorder

- Dietary deficiency
- Certain drugs and poisons
- Unknown factors.

Classifications of neuromuscular disorders

Some of the major diseases which affect the neuromuscular system are classified into four main groups, including:

- **Motor neurone diseases** – for unknown or genetic reasons, the lower (and sometimes also the upper) motor neurons gradually die. Some of the different types of genetic (inherited) motor neuron diseases include infantile progressive spinal muscular atrophy (SMA1), intermediate spinal muscular atrophy (SMA2), juvenile spinal muscular atrophy (SMA3) and adult spinal muscular atrophy. The most common form of motor neuron disease, known simply as motor neurone disease or amyotrophic lateral sclerosis or Lou Gehrig's disease, is usually not inherited and its cause remains unknown.
- **Neuropathies** – the peripheral nervous system (nerves other than those within the spinal cord) are affected. Some of the different diseases of the peripheral nerve include the genetic disease Charcot-Marie-Tooth disease, the hormonal disorder diabetes (if poorly controlled), and autoimmune diseases such as chronic inflammatory demyelinating neuropathy (CIDP).
- **Neuromuscular junction disorders** – in these diseases, the transmission of the signal to move (contract) a muscle is blocked as it tries to bridge the gap between the nerve and muscle. The most common of these diseases is myasthenia gravis, an autoimmune disease where the immune system produces antibodies that attach themselves to the neuromuscular junction and prevent transmission of the nerve impulse to the muscle.
- **Myopathies including muscular dystrophies** – many different types of muscular dystrophy (muscle wastage) are caused by various genetic mutations that prevent the maintenance and repair of muscle tissue. Some of the different types include Becker muscular dystrophy, congenital muscular dystrophy, Duchenne muscular dystrophy and facioscapulohumeral muscular dystrophy. Other diseases of the muscles (myopathies) can be caused as a rare side effect of medications (for example, the cholesterol-lowering drugs known as statins), autoimmune disease such as polymyositis or polymyalgia rheumatica or hormonal disorders such as hypothyroidism.

Diagnosis and treatment of neuromuscular disorders

Depending on the condition, neuromuscular disorders are diagnosed using a range of tests, including electrical tests known as nerve conduction studies (to measure the ability of nerves to conduct electricity), electromyography (EMG) to examine the health of a muscle, blood tests, muscle biopsies and genetic testing.

Treatment varies widely, according to the individual disorder, and some conditions are more easily treated than others.

Where to get help

- Your doctor
- Muscular Dystrophy Association Australia Tel. (03) 9320 9555

Things to remember

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This page has been produced in consultation with and approved by:

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