Myoclonus

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What is myoclonus?

Myoclonus refers to sudden, brief involuntary twitching or jerking of a muscle or group of muscles. The twitching cannot be stopped or controlled by the person experiencing it. Myoclonus is not a disease itself, rather it describes a clinical sign.

Myoclonic jerks may occur in the following scenarios:

- Either alone or in sequence, in a pattern of movement or without pattern
- Infrequently or many times per minute
- In response to an external event or when a person attempts to make a movement

Types of myoclonus

Myoclonus can be broadly categorized into two types:

1. Physiologic myoclonus involves quick muscle twitches followed by relaxation. Examples are hiccups and the jerks or "sleep starts" that some people experience while drifting off to sleep. This form occurs in healthy people, causes no difficulties, and does not require medical treatment.
2. Pathologic myoclonus may involve persistent, shock-like contractions in a group of muscles and is more widespread in general. They contractions begin in one region of the body and spread to muscles in other areas. More severe cases can affect movement and severely limit a person's ability to eat, talk, or walk. This can be one of many signs indicating a wide variety of underlying disorders in the brain or nerves, secondary to certain medical conditions, or can be a reaction to certain types of medication.

Classifying myoclonus is difficult because the causes and responses to therapy vary widely. Some of the commonly described types are:

- Stimulus-sensitive myoclonus, which is triggered by various external events, including noise, movement, and light. Being surprised may increase the sensitivity of the individual.
- Sleep myoclonus (also known as hypnic myoclonus) occurs during sleep and sleep transitions, often as one is drifting off to sleep. Some forms appear to be stimulus sensitive. While some people may not be troubled by this or need treatment, others may require treatment where myoclonus may be a symptom of a more complex and disturbing sleep disorder.
- Essential myoclonus occurs on its own and is not influenced by abnormalities in the brain or nerves. Involuntary twitches or spasms can occur in people with no family history of the condition, and the cause may be unexplained (idiopathic). However, it also can appear among members of the same family—indicating that it may be an inherited disorder. It tends to be stable without increasing in severity over time. In some families there is an association of essential myoclonus with essential tremor or a form of dystonia (myoclonus-dystonia). Dystonia is a movement disorder in which sustained muscle contractions cause twisting and repetitive movements or uncomfortable postures.
- Action myoclonus is triggered by voluntary movement or even the intention to move. It may become worse during attempts at precise, coordinated movements. It can be the most disabling form of myoclonus affecting the arms, legs, and face. One of the causes may be brain damage that results from a lack of oxygen and blood flow to the brain, or it can be secondary to other medical or neurological conditions.
- Cortical reflex myoclonus originates in the cerebral cortex (the outer layer of the brain that is largely responsible for information processing). Myoclonic jerks usually involve only a few muscles in one part of the body, but jerks involving many muscles also may occur. It becomes more intense when a person attempts to move in a certain way (action myoclonus) or perceives a particular sensation.
- Epileptic myoclonus is the presence of myoclonus in people living with epilepsy. Myoclonus can occur as the only seizure manifestation, as one component of a seizure, or one of multiple types of seizures within an epilepsy syndrome. Some examples of syndromes with myoclonic seizures include:
    - Juvenile myoclonic epilepsy (JME) starts around puberty and involves myoclonic seizures usually of the neck, shoulders, or upper arms, as well as generalized tonic-clonic seizures (affecting the whole body).
    - Myoclonic-astatic epilepsy has generalized myoclonic jerks or seizures followed by a loss of muscle tone.
    - Lennox-Gastaut Syndrome occurs in childhood and involves multiple seizure types that are usually difficult to control as well as cognitive impairment.
    - Progressive myoclonus epilepsy (PME) is a group of rare disorders characterized by myoclonic seizures and other neurologic symptoms such as trouble walking or speaking. These disorders often get worse over time and sometimes are fatal. One of its many forms is Lafora body disease (also known as Lafora progressive myoclonus epilepsy), which is characterized by myoclonic seizures, progressive loss of memory, and impaired intellectual functions.
- Reticular reflex myoclonus originates in the brain stem (the part of the brain that connects to the spinal cord and controls vital functions such as breathing and heartbeat). Myoclonic jerks usually affect the whole body, with muscles on both sides of the body affected simultaneously. In some people, myoclonic jerks occur in only one part of the body, such as the legs, with all the muscles in that part being involved in each jerk. It can be triggered by either a voluntary movement or an external stimulus.
- Palatal myoclonus (also known as palatal tremor) is a regular, rhythmic contraction of one or both sides of the rear of the roof of the mouth (soft palate). The contractions are rapid and may continue during sleep. The condition usually appears in adults and can last indefinitely. People with palatal myoclonus may note a “clicking” sound in the ear when the muscles in the soft palate contract. This can be idiopathic or secondary to injury in the brain stem or adjacent cerebellum.
- Spinal myoclonus originates in the spinal cord. In some instances, the myoclonic jerk

involves the whole trunk of the body, beginning in the thoracic (middle) spinal segments and spreading up and down, a phenomenon known as propriospinal myoclonus.
- Peripheral myoclonus refers to myoclonic jerks that originate from a peripheral nerve (outside of the brain and spinal cord) such as in hemifacial spasm (frequent spasms of the muscles on one side of the face).

Who is more likely to get myoclonus?

Myoclonus may be caused by the following:

- A disturbance of the brain or spinal cord (the central nervous system, or CNS)—most common
- An injury to the peripheral nerves (the nerves outside the CNS that connect to sensory organs and muscles, and relay information from/to the CNS)—least common

Myoclonus can occur by itself or as one of several symptoms associated with a wide variety of nervous system disorders. For example, myoclonic jerks may develop in individuals with multiple sclerosis or epilepsy, and with neurodegenerative diseases such as Parkinson's disease, Alzheimer's disease, or Creutzfeldt-Jakob disease.

Myoclonus may also be seen in conjunction with infection, head or spinal cord injury, stroke, brain tumors, kidney or liver failure, chemical or drug intoxication, or metabolic disorders. Prolonged oxygen deprivation to the brain (hypoxia) may lead to post-hypoxic myoclonus.

Myoclonus can begin in childhood or adulthood with symptoms ranging from mild to severe.

Myoclonic twitches or jerks are caused by:

- Sudden muscle contractions (tightening), called positive myoclonus
- Muscle relaxation, called negative myoclonus

Studies suggest that the following locations in the brain are involved in myoclonus:

- Cerebral cortex, the most common origin for myoclonus
- Brain stem, close to structures that are responsible for the startle response, which is an automatic reaction to an unexpected stimulus involving rapid muscle contraction

However, the specific mechanisms underlying myoclonus are not yet fully understood. Scientists believe that some types of stimulus-sensitive myoclonus may involve overexcitability of the parts of the brain that control movement. Laboratory studies suggest that an imbalance between chemicals called neurotransmitters may bring about myoclonus with the end result being a lack of inhibition at some level (inhibition is a decrease in the rate of a chemical reaction or its prevention).

Neurotransmitters carry messages between nerve cells. They are released by one nerve cell and attach to a protein called a receptor on neighboring (receiving) cells. Abnormalities or deficiencies in receptors for certain neurotransmitters may contribute to some forms of myoclonus, including receptors for:

- Serotonin, which is involved in managing mood, cognition, reward, learning, memory, physiological processes, etc.
- Gamma-aminobutyric acid (GABA), which is involved in motor control
- Glycine, which is important for the control of motor and sensory functions in the spinal cord
- Opioids, which are involved in different functions related to analgesia, pain, and depression

More research is needed to determine how these receptor abnormalities cause or

contribute to myoclonus.

How is myoclonus diagnosed and treated?

Diagnosing myoclonus

Following a review of your medical history and physical exam, a doctor may order additional tests to confirm a diagnosis of myoclonus:

- Electromyography (EMG) to measure electrical activity of muscle
- Electroencephalography (EEG) to record the electrical activity of the brain that may trigger the myoclonic jerk through electrodes attached to the scalp
- Evoked potential studies to capture the electrical activity in the brain, brain stem, and spinal cord evoked by specific stimuli (tactile, auditory, visual stimulation)
- Laboratory urine or blood tests to look at possible causes and to rule out other conditions that may cause symptoms similar to myoclonus
- Magnetic resonance imaging (MRI) to produce three-dimensional images of the brain, spinal cord, nerve, and other tissue (including muscles).

Treating myoclonus

A doctor's first consideration in treating myoclonus is reversing or treating any underlying cause or the origin of the myoclonus. However, many cases require symptomatic treatment if the myoclonus is disabling.

Several options are available to help treat myoclonus:

- Clonazepam, a type of tranquilizer, is the most commonly used medication to treat some forms of myoclonus.
- Other drugs such as certain barbiturates, phenytoin, levetiracetam, valproate, and primidone are used to treat epilepsy in addition to myoclonus.
- Multiple medications may be required for effective treatment because some medications have a limited effect when used individually but may have a greater effect when combined with others.
- Hormonal therapy may improve responses to antimyoclonic drugs in some people.
- 5-hydroxytryptophan (5-HTP), a building block of serotonin (a chemical made in the body that transmits nerve impulses), leads to improvement in individuals with some types of action myoclonus and progressive myoclonus epilepsy. However, the effectiveness of 5-HTP therapy varies between individuals, and can make the condition worse in some individuals.
- Botulinum toxin injections can reduce excess muscle activity by blocking the activity of a chemical that makes muscles contract at the cellular level. It is the first-line therapy for hemifacial spasm (frequent spasms of the muscles on one side of the face) and has been effective in treating some individuals with palatal myoclonus.

What are the latest updates on myoclonus?

The National Institute of Neurological Disorders and Stroke (NINDS), a component of the National Institutes of Health (NIH), supports research on myoclonus at its laboratories in Bethesda, Maryland and through grants to major research institutions across the country. These research projects include:

- Biomarkers are measurable indicators of some biological state or condition and are often necessary for predicting the nature and severity of a disease. The Juvenile Myoclonic Epilepsy Connectome Project (JMECP) aims to define biomarkers of JME. Using state-of-the-art imaging methods, NINDS-funded researchers measure altered structural and functional connections between brain regions in children and adolescents between 12 to 20 years of age who are living with JME. Results may lead to novel clinical tools for diagnosis and personalized management.
- Glycogen is a form of sugar that is used as an energy reserve in many cells. Lafora bodies (LBs) are unusual, glycogen-like inclusions found in cells of all tissues in individuals suffering from Lafora body disease (or Lafora progressive myoclonus epilepsy). In another study, NINDS-funded researchers generate proteins that will help to degrade or break down LBs, which could lead to a novel therapeutic strategy to treat Lafora body disease.
- Researchers of the Lafora Epilepsy Cure Initiative determined the satisfactory performance of therapeutic agents against Lafora body disease and myoclonus epilepsy in pre-clinical trials with mice. A current NINDS-funded study will develop an Early Diagnosis Campaign to prepare a clinical trial ready group of people with early-stage and moderately advanced Lafora body disease and to identify clinical biomarkers of disease progression before advancing from mice therapeutics to human clinical trials.
- Botulinum toxin is a treatment for a variety of movement disorders. A NINDS study compared the use of ultrasound (using sound waves) and electrophysiologic guidance (using electrical stimulation and a needle) to precisely target muscles for botulinum toxin injection to treat upper limb spasticity and focal hand dystonia. Results may lead to improved treatment for movement disorders like myoclonus.
- Animal models are used to study the mechanisms involved in myoclonus. For example, NINDS-funded scientists have developed a mouse model of myoclonus-dystonia (an inherited movement disorder characterized predominantly by myoclonus of the upper body and dystonia). A striking characteristic of this disorder is that motor symptoms improve with alcohol consumption. Researchers tested the hypothesis that abnormal activity of the cerebellum (the part of the brain responsible for coordination and regulation of voluntary movement) causes myoclonus and dystonia in myoclonus-dystonia, and that by acting on targets in the cerebellum, alcohol injections normalize cerebellar activity to relieve motor symptoms. Results may provide a better understanding of the underlying neurological cause of myoclonus and dystonia in myoclonus-dystonia and provide targets for treatment options.
- Complex movement disorders (CMDs), or disorders in which individuals are affected by more than one movement disorder, such as parkinsonism and dystonia, or myoclonus and tremor, are a continuing challenge for diagnosis and treatment. NINDS-funded researchers are recruiting individuals with familial and sporadic CMDs to identify genetic mutations that may cause these disorders. Findings may lead to improvements in disease diagnosis and treatment.

In addition to NINDS, other NIH institutes and centers support research on movement disorders that include myoclonus. More information is available through the NIH RePORTER, a searchable database of current and previously funded research, as well as research results and publications.

For research articles and summaries on myoclonus, search PubMed, which contains citations from medical journals and other sites.

Learn About Clinical Trials

Clinical trials are studies that allow us to learn more about disorders and improve care.

They can help connect patients with new and upcoming treatment options.

Search Clinical Trials

How can I or my loved one help improve care for people with myoclonus?

Consider participating in a clinical trial so clinicians and scientists can learn more about myoclonus. Clinical research uses human volunteers to help researchers learn more about a disorder and perhaps find better ways to safely detect, treat, or prevent disease.

All types of volunteers are needed—those who are healthy or may have an illness or disease—of all different ages, sexes, races, and ethnicities to ensure that study results apply to as many people as possible, and that treatments will be safe and effective for everyone who will use them.

For information about participating in clinical research visit NIH Clinical Research Trials and You. Learn about clinical trials currently looking for people with myoclonus at Clinicaltrals.gov, a searchable database of current and past federal and private clinical studies.

Where can I find more information about myoclonus?

Information may be available from the following resources:

MedlinePlus

National Organization for Rare Disorders (NORD)
Phone: 800-999-6673

Learn about related topics

- Alzheimer's Disease
- Brain and Spinal Cord Tumors
- Creutzfeldt-Jakob Disease
- Epilepsy and Seizures
- Multiple Sclerosis
- Parkinson's Disease
- Spinal Cord Injury
- Stroke
- Traumatic Brain Injury

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