Other Diseases Affecting Muscle Membrane Excitability

What?

Question 1

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A. Lumbar Puncture

B. MRI of the lumbosacral plexus

C. Muscle Biopsy

D. No further testing is needed; refer to PT

E. Trial of IV methylprednisolone

Question 2

A 45 year old man presents with 2 years of muscle twitches and cramps in all 4 extremities, calf muscle hypertrophy, hyperhidrosis, normal strength, normal reflexes, and normal sensation. He has normal facial features. His cognition and sleep are normal. EMG shows myokymic discharges and neuromyotonia. Which disorder is most likely?

A. Morvan Syndrome

B. Isaac's syndrome

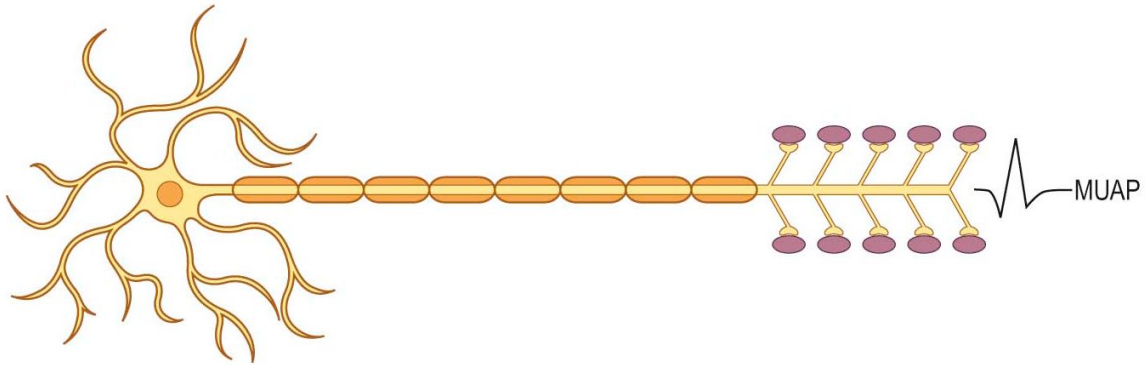
C. Stiff Person Syndrome

D. Cramp-Fasciculation Syndrome

E. Myotonic dystrophy type I

States of Persistent Muscle Activity

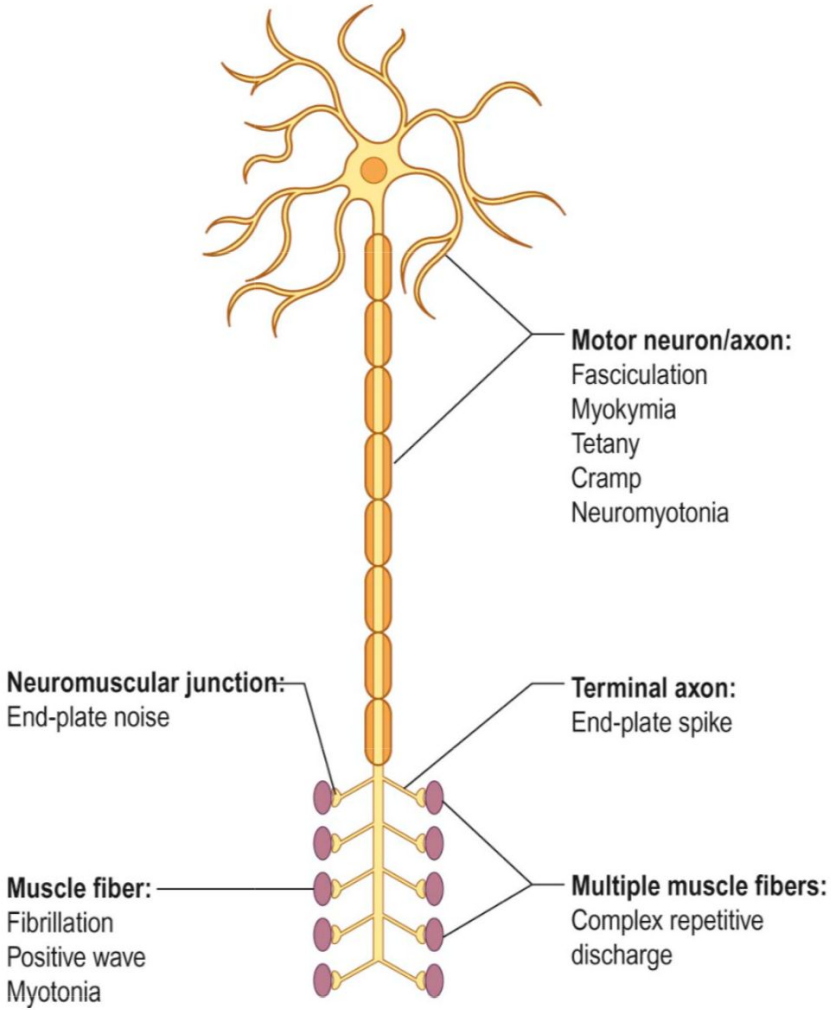
Muscles at rest should be quiet



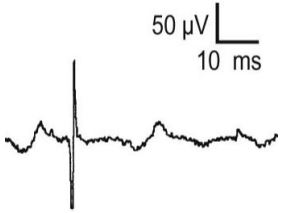
Motor Unit: Anterior horn cell, axon, and all the muscle fibers with which it is connected

Injured Motor Units Lead to Spontaneous Complaints

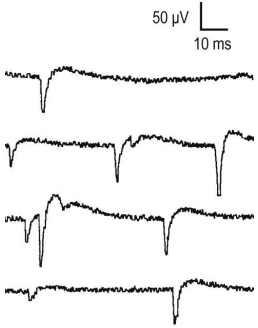
- Abnormal spontaneous activity



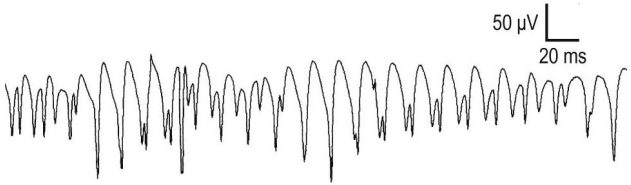
Spontaneous Activity: Abnormal Muscle Fiber Potentials



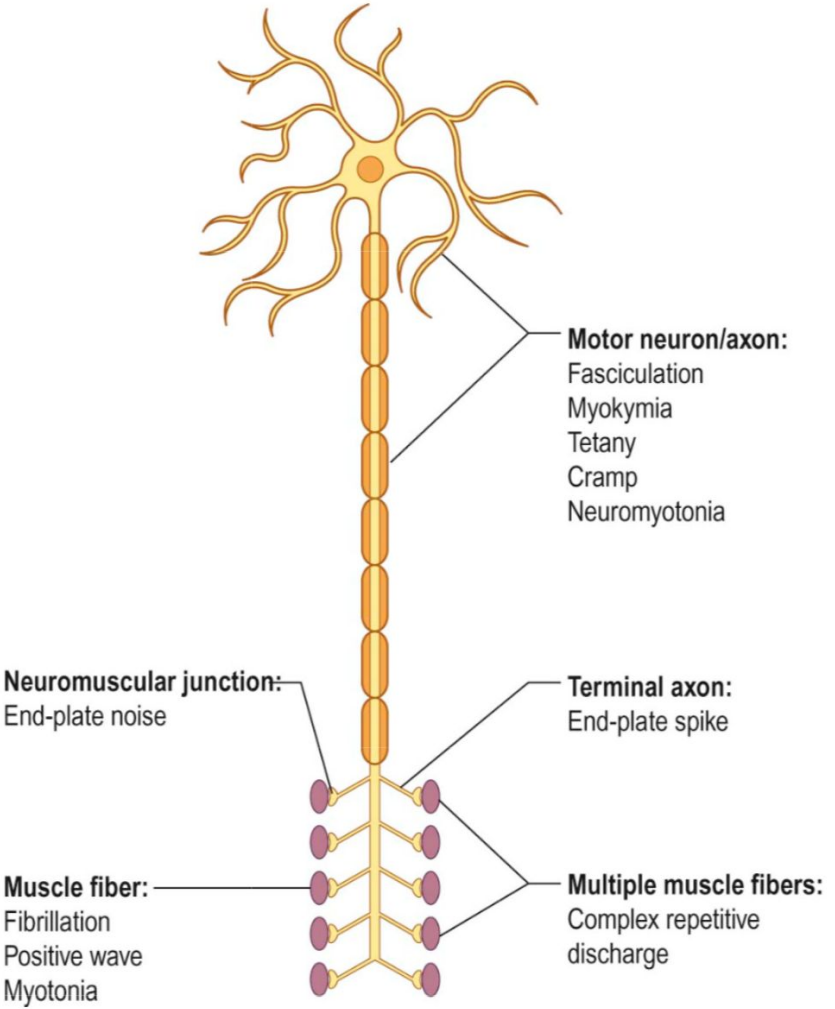
Fibrillation



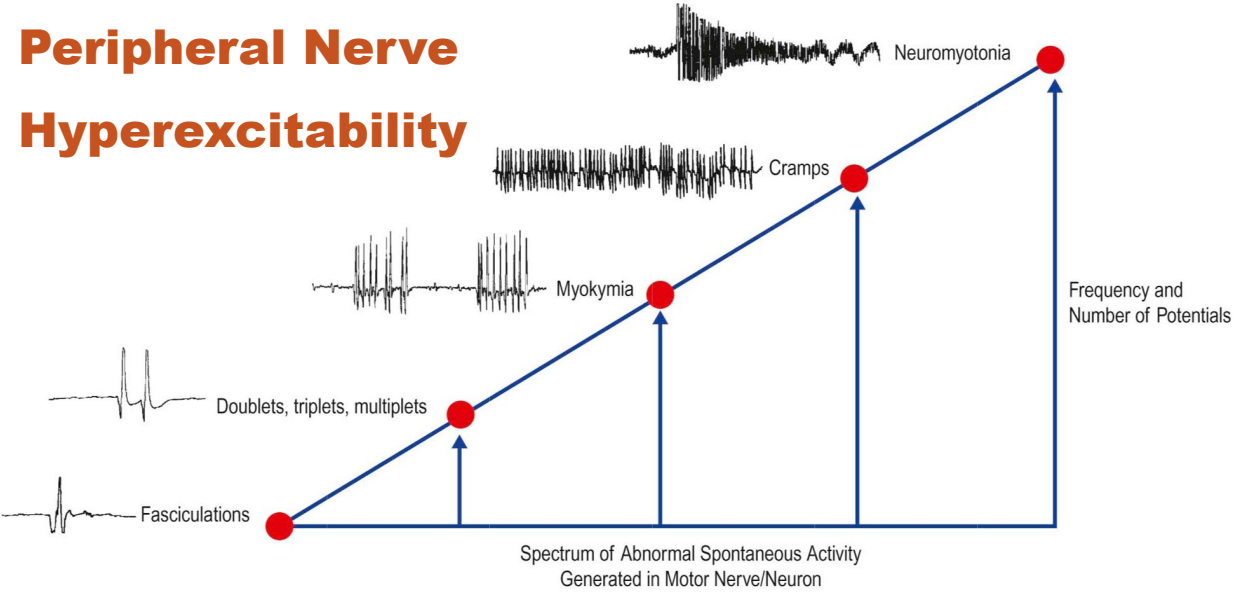
Positive Sharp Waves



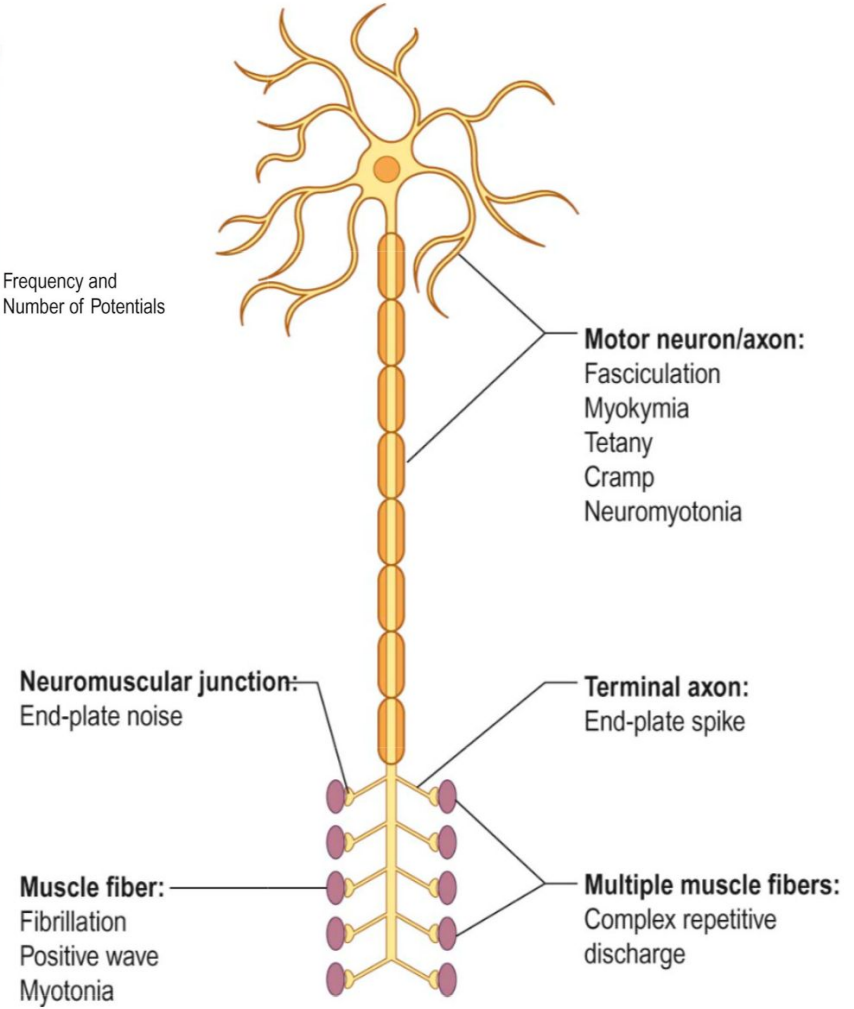
Myotonic Discharges



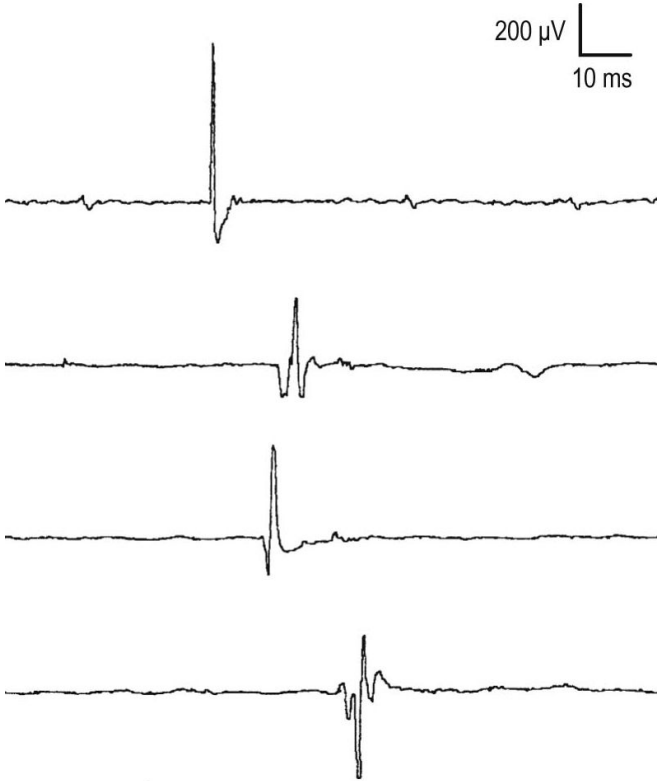
Peripheral Nerve Hyperexcitability



Spontaneous Activity: Abnormal Motor Unit (Motor Neuron/Axon) Potentials



Fasciculations



- Single spontaneous involuntary discharge of a motor unit
- Slow (less than 1-2 Hz)
- Look like a motor unit (notice size compared to fib/psw)
- Seen in any process of lower motor neuron, and can be seen in normal people
- Sound like corn popping
- Brief twitch without movement of a joint

Fasciculations

- RUSH Fasciculation video
  - https://youtu.be/Q_DrreXa-5Q

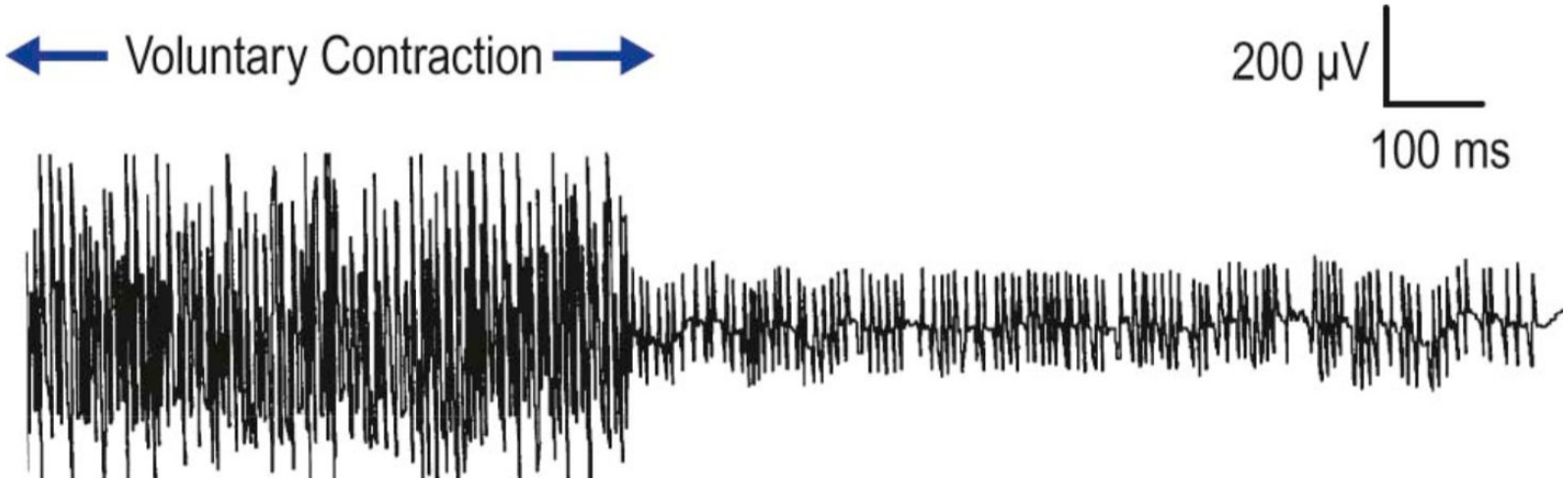
Benign Fasciculation Syndrome

- Few random fasciculations are normal
- Rule of thumb: fasciculations without weakness, atrophy, or reflex change is not indicative of motor system disease
- Commonly seen in thenar eminence, eyelids, calves, or orbicularis oculi
- Benign fasciculations tend to be more frequent and rhythmic and affect the same site repetitively
- Commonly seen in physicians or family members of those with ALS
- BFS doesn’t progress, though infrequently patients may initially be thought to have BFS, who later develop ALS
- Carbamazepine may help

Cramp Fasciculation Syndrome

- Variant of BFS, with conjoined cramps, stiffness, exercise intolerance
- Cramps are high-frequency discharges of motor axons, not muscle
- May be disabling to some degree, but prognosis is good
- Salient feature: Stimulation of peripheral nerves results in sustained muscle firing due to prolonged trains of action potentials in the distal motor nerve
- Small number of these patients have autoantibodies against voltage-gated potassium channels
- Carbamazepine may help
- Cramps may also be seen in lots of disorders (neuropathic, endocrine, metabolic), and can be benign (nocturnal calf cramps)

Cramp Fasciculation Syndrome



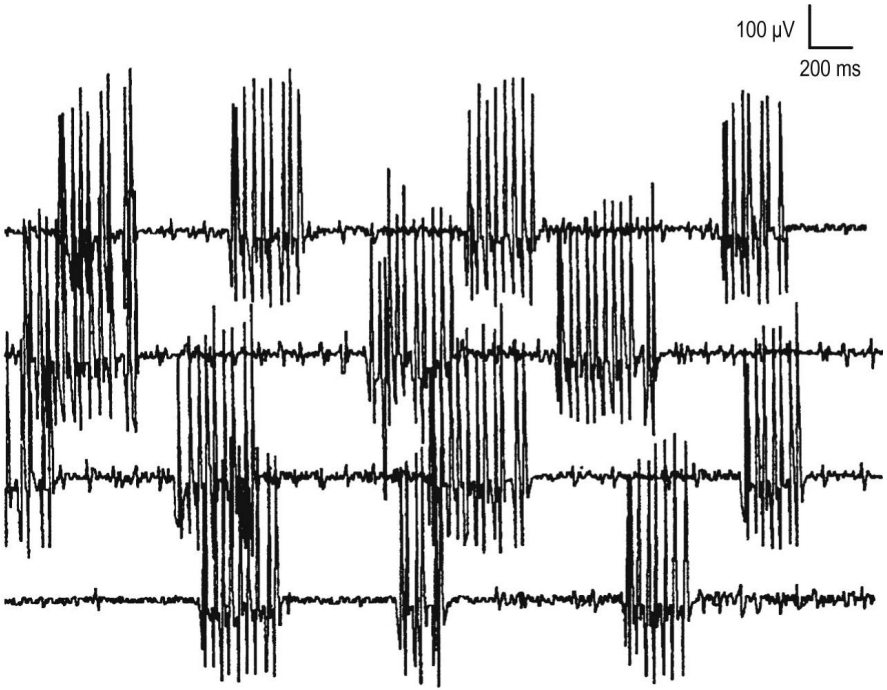
Myokymia

- State of abnormal rippling muscle activity
- Rhythmic group of spontaneous repetitive discharges of the same motor unit (grouped fasciculations)
- Marching sound on EMG (irregular marching steps)
- Clinically continuous involuntary quivering, rippling, undulating movement
- Generalized or limited to one part of the body
- Neuropathic
  - May be seen in Guillain-Barre syndrome, MS, pontine tumor affecting the facial nerve
  - BOARD QUESTION: Radiation damage to the brachial plexus or lumbar plexus.
  - May also occur in radics, neuropathies, and even spinal cord lesion with demyelination

Myokymia

Youtube video of bifacial myokymia with brainstem glioma

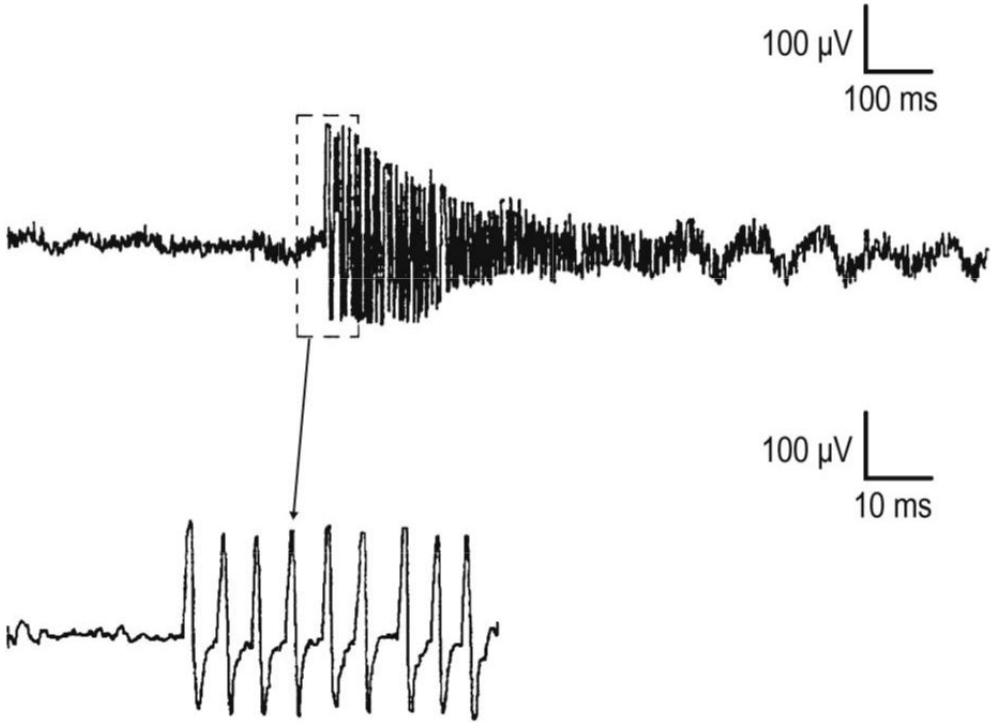
- https://youtu.be/GAWTuBYf2bk



Neuromyotonia

- High frequency (highest frequency of any discharge) decrementing repetitive discharges of a single motor unit, rare
- “Ping sound” on EMG
- Clinically display generalized stiffness, hyperhidrosis, delayed muscle relaxation after contraction (not elicited by muscle percussion, unlike myotonia)
- Myotonia=discharge of muscle fiber. Neuromyotonia=motor neuron discharge
- **Isaac’s Syndrome**= Acquired neuromyotonia, can be autoimmune disorder (Voltage-gated K channel Ab, Caspr2 Ab, LFI1 Ab), paraneoplastic (thymoma, SCLC, Hodgkin lymphoma) continuous muscle twitching and myokymia, muscle hypertrophy, weight loss, and hyperhidrosis
- **Morvan’s Syndrome**= Isaac’s plus encephalopathy, HA, drowsiness, hallucinations, seizures, sleep alterations, autonomic dysfunction. Also with VGKC Abs, Caspr2 Ab, thymoma.

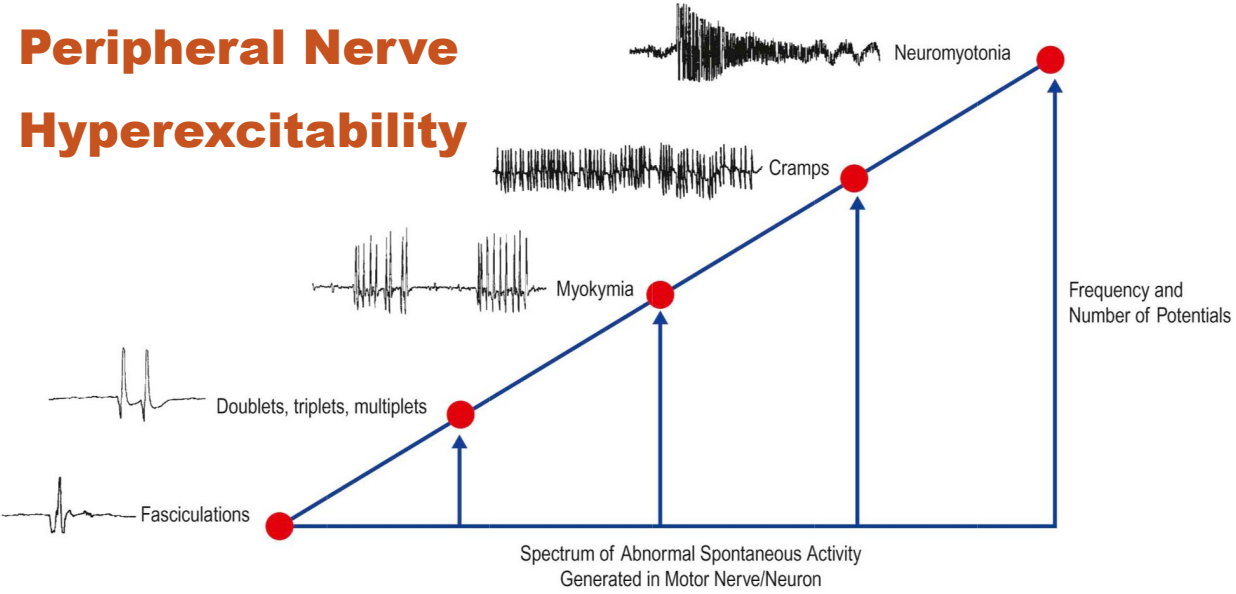
Neuromyotonia



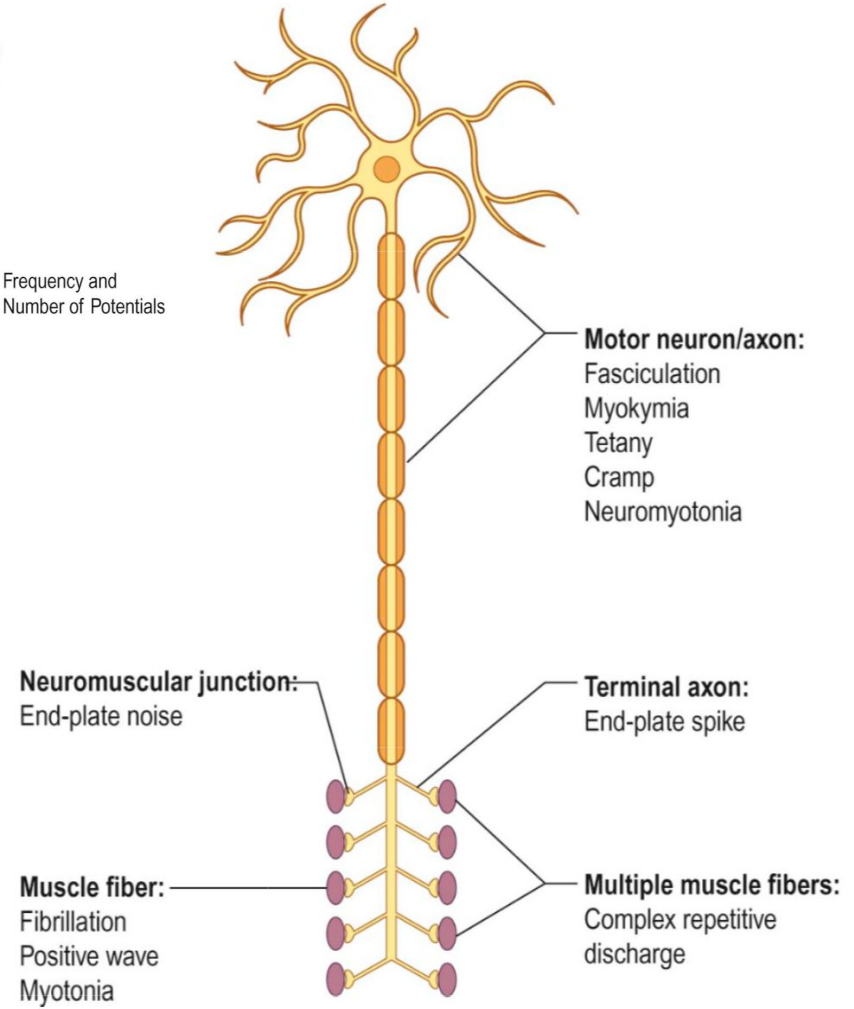
Other Diseases of Muscle Spasms (not due to motor neuron pathology)

- Tetanus= Toxin inhibits the inhibition of spinal and brainstem motor neurons typically provided by the Renshaw cells (in the **cord**), resulting in involuntary muscle spasms.
- Stiff Person Syndrome= Spasm and stiffness result of disinhibition of interneurons in the gray matter of the **spinal cord**. This is caused by GAD Abs (GAD is the synthesizing enzyme for GABA, which is supposed to inhibit input to motor neurons).
- Black Widow Spider= Toxin triggers rapid presynaptic release of acetylcholine triggering muscle spasms and pain.

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References

Victor and Adams - Principles of Neurology
Pages 1494-1499

Preston and Shapiro - EMG and Neuromuscular Disorders

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