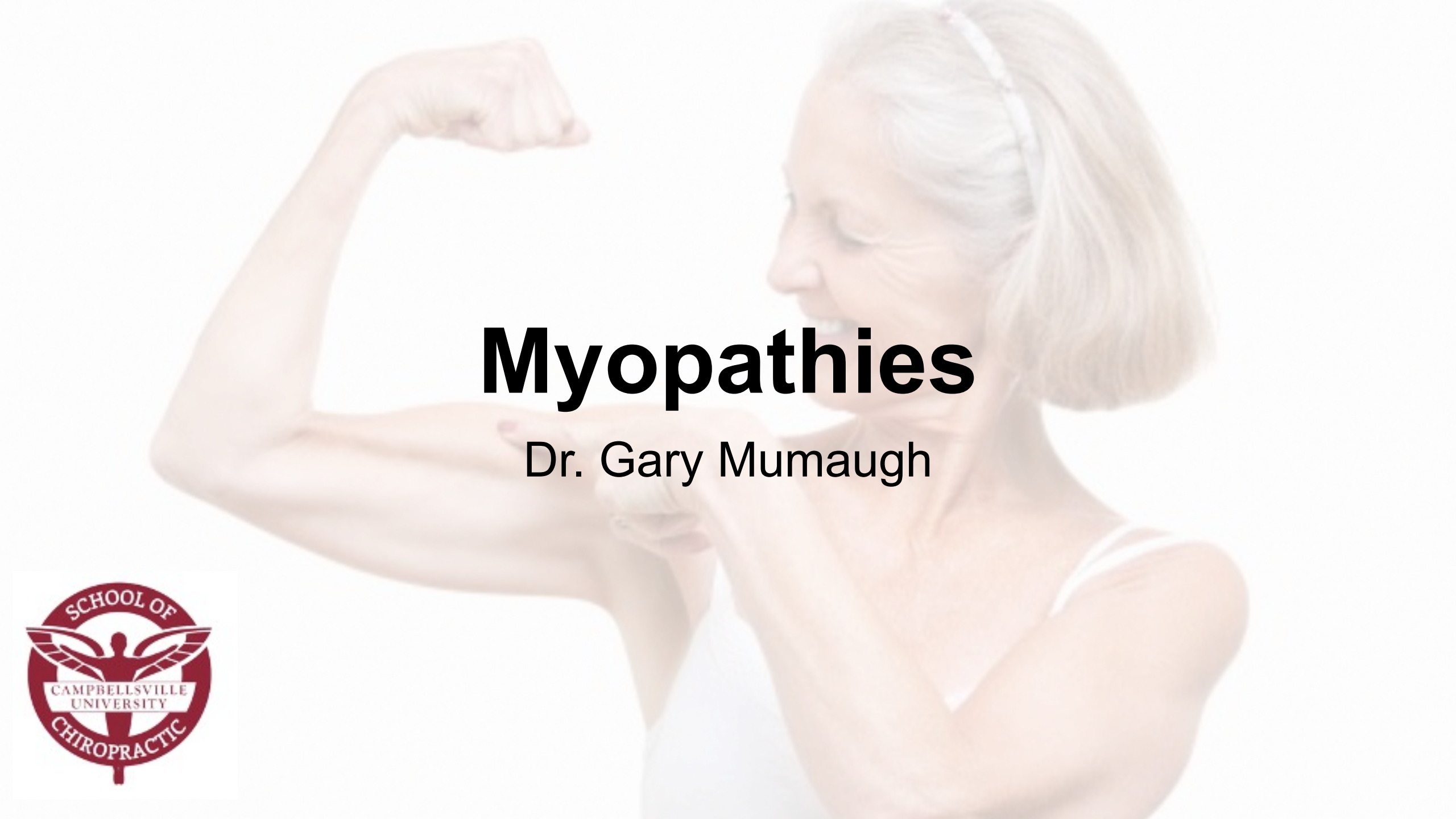


Myopathies

Dr. Gary Mumaugh

SCHOOL OF CHIROPRACTIC
CAMPBELLSVILLE UNIVERSITY

Disease Categories in Neurology

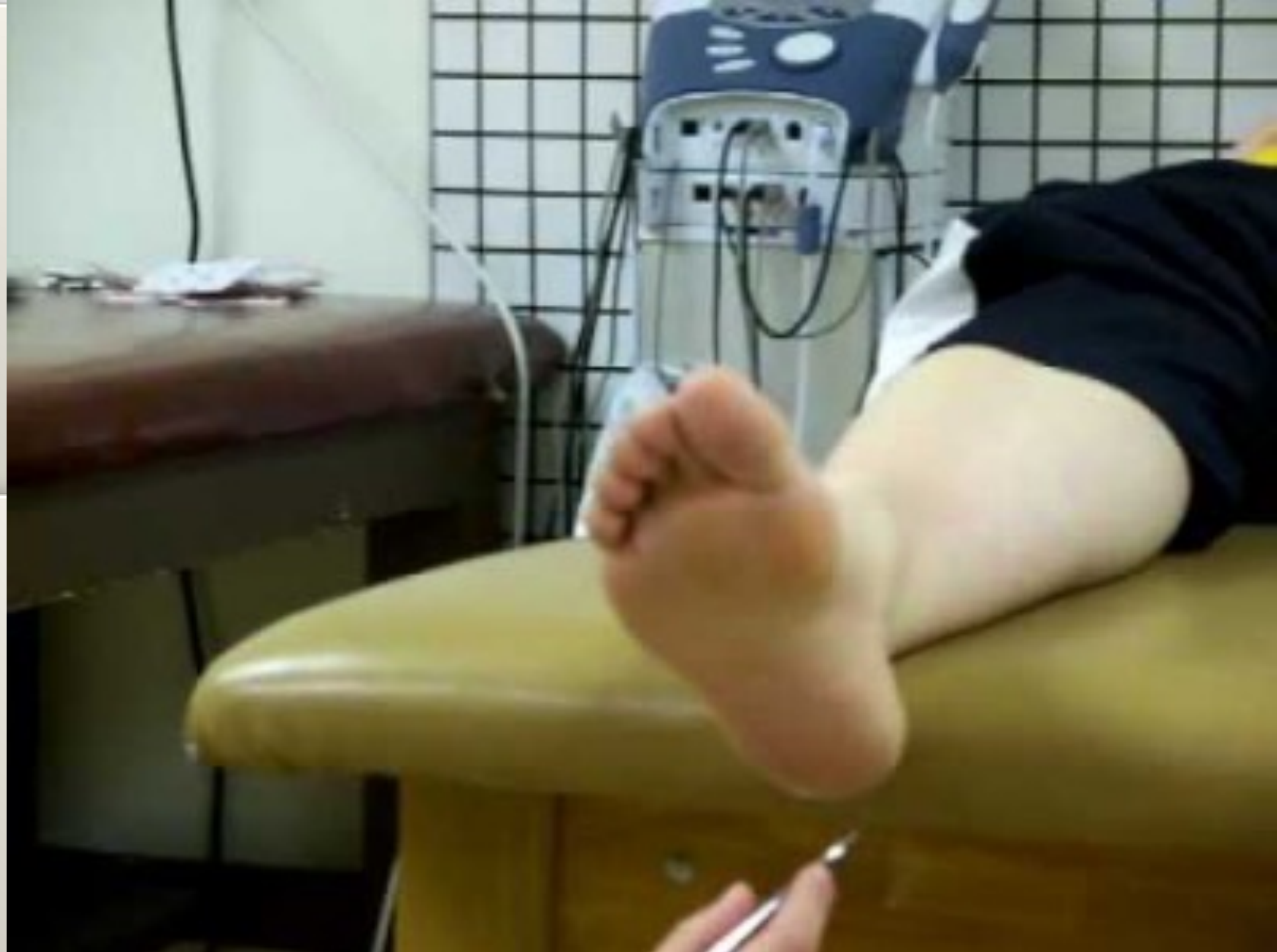
- Myopathy – muscle diseases
- Myelopathy – cord compression diseases
- Neuropathy – nerve diseases
- Radiculopathy – nerve root compression
- Plexopathy – nerve plexus compression
- Encephalopathy – brain diseases

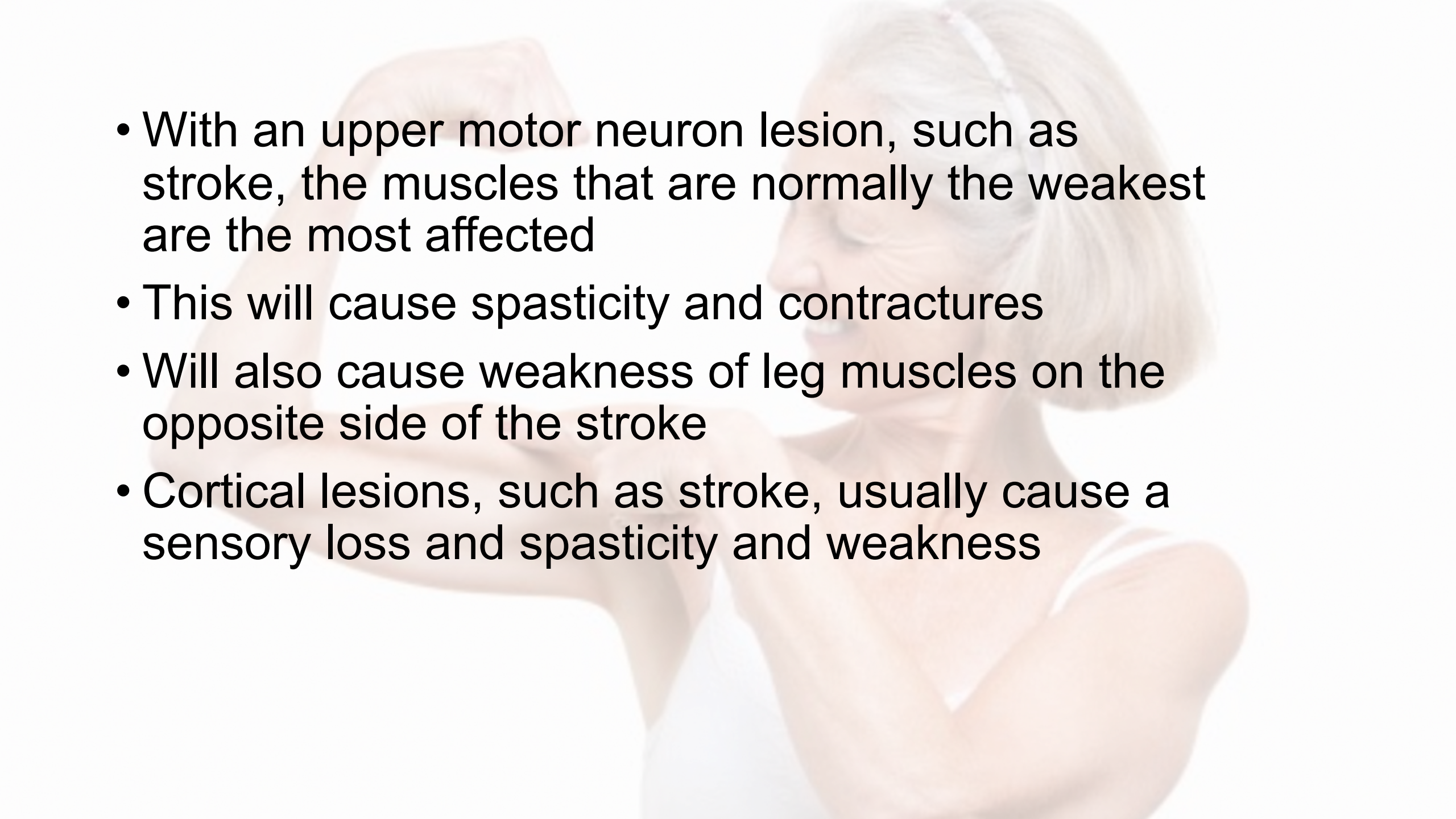
Weakness Disorders

- Can range from mild weakness to total paralysis
  - Ask “Is this specific muscle fatigue or weakness or are all muscles weak?”
    - “If all muscles are weak, is it muscular, exhaustion or neurological?”
    - “Is it only one side of the body?”
    - “Is it upper motor neuron problem or lower motor neuron problem?”

Upper Motor Neuron Problems

- An upper motor neuron lesion is a lesion of the neural pathway above the anterior horn cell or motor nuclei of the cranial nerves
- Spasticity, increase in tone in the extensor muscles (lower limbs) or flexor muscles (upper limbs)
- Weakness in the flexors (lower limbs) or extensors (upper limbs), but no muscle wasting
- Babinski sign is present, where the big toe is raised (extended) rather than curled downwards (flexed) upon appropriate stimulation of the sole of the foot. The presence of the Babinski sign is an abnormal response in adulthood
- Increase Deep tendon reflex (DTR)





- With an upper motor neuron lesion, such as stroke, the muscles that are normally the weakest are the most affected
- This will cause spasticity and contractures
- Will also cause weakness of leg muscles on the opposite side of the stroke
- Cortical lesions, such as stroke, usually cause a sensory loss and spasticity and weakness

Lower Motor Neuron Lesion

- A lower motor neuron lesion is a lesion which affects nerve fibers traveling from the anterior horn of the spinal cord to the relevant muscle(s) -- the lower motor neuron
- One major characteristic used to identify a lower motor neuron lesion is flaccid paralysis - paralysis accompanied by muscle loss.
- This is in contrast to a upper motor neuron lesion, which often presents with spastic paralysis- paralysis accompanied by severe hypertonia
- An example of a lower motor neuron lesion is an ulnar nerve neuropathy

UMN Lesion vs LMN Lesion

SIGN	UMN	LMN
Weakness	Yes	Yes
Atrophy	No *	Yes
Fasciculations	No	Yes
Reflexes	Increased	Decreased
Tone	Increased	Decreased

* May have mild atrophy due to disuse

Neuropathy

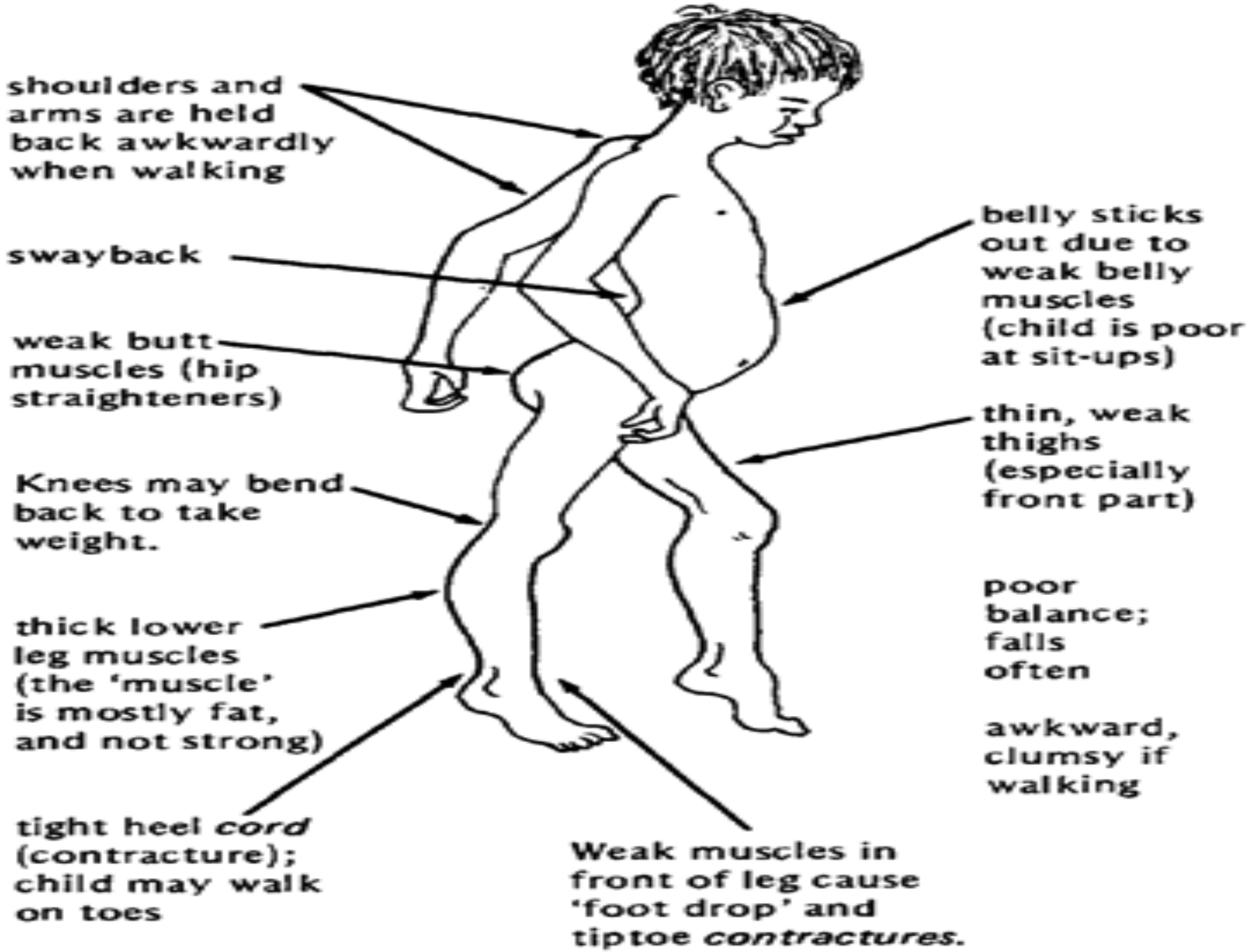
- Affects distal nerves in a glove like pattern
- Paresthesias, weakness, sensory loss
- Common in diabetes, RA, alcoholic abuse and B12 deficiency

Disease of muscle - myopathy

- These diseases cause proximal muscle weakness
- Classic disease is muscular dystrophy
  - Usually affects young boys with weakness of pelvic and shoulder muscles
  - The affected muscles are large and bulky, but very weak because the muscle cells do not contract properly

Muscular Dystrophy - a mysterious disease

Muscular Dystrophy Walking

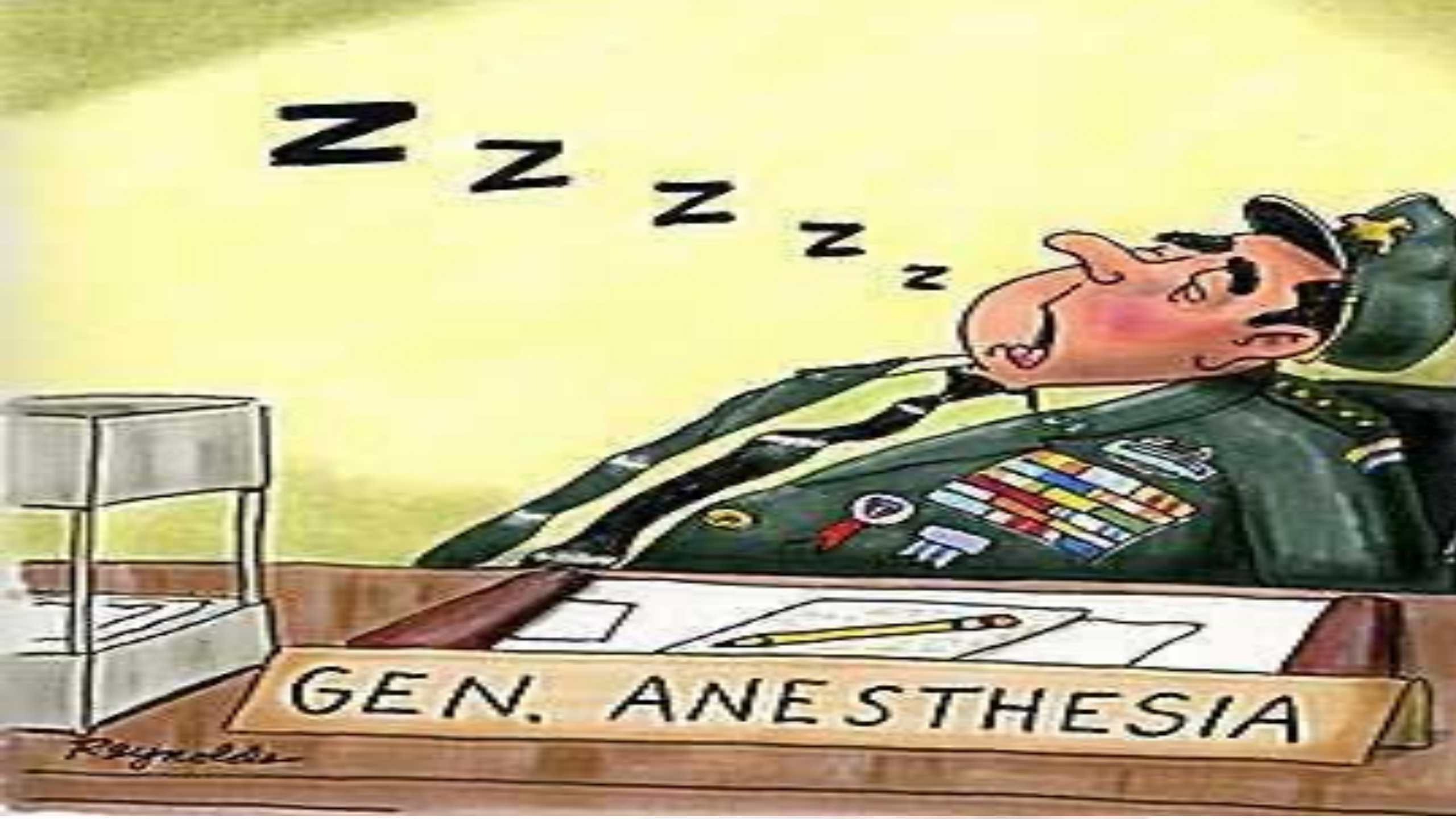

shoulders and arms are held back awkwardly when walking
swayback
weak butt muscles (hip straighteners)
Knees may bend back to take weight.
thick lower leg muscles (the 'muscle' is mostly fat, and not strong)
tight heel *cord* (contracture); child may walk on toes
Weak muscles in front of leg cause 'foot drop' and tiptoe *contractures*.
belly sticks out due to weak belly muscles (child is poor at sit-ups)
thin, weak thighs (especially front part)
poor balance; falls often
awkward, clumsy if walking

Pain & Sensory Loss Syndromes

- Pain syndromes
  - Neuritis or neuritic pain – pain due to nerve dysfunction can be very severe pain
  - Example is causalgia which comes on months after a crushing extremity injury
  - The resulting pain is so severe that patient's will often request amputation to relieve the pain
- Sensory loss – can cause four things:
  - Anesthesia – loss of sensation
  - Hypoesthesia – decreased sensation
  - Paresthesia –numbness, tingling, prickly
  - Dysesthesia – uncomfortable burning sensation

"You can have general anesthesia or just be numbed from the wallet down."

"OK, Bernice. He no longer seems to feel your punches. The Novocain seems to have kicked in."


Z z z z
GEN. ANESTHESIA
Reynolds

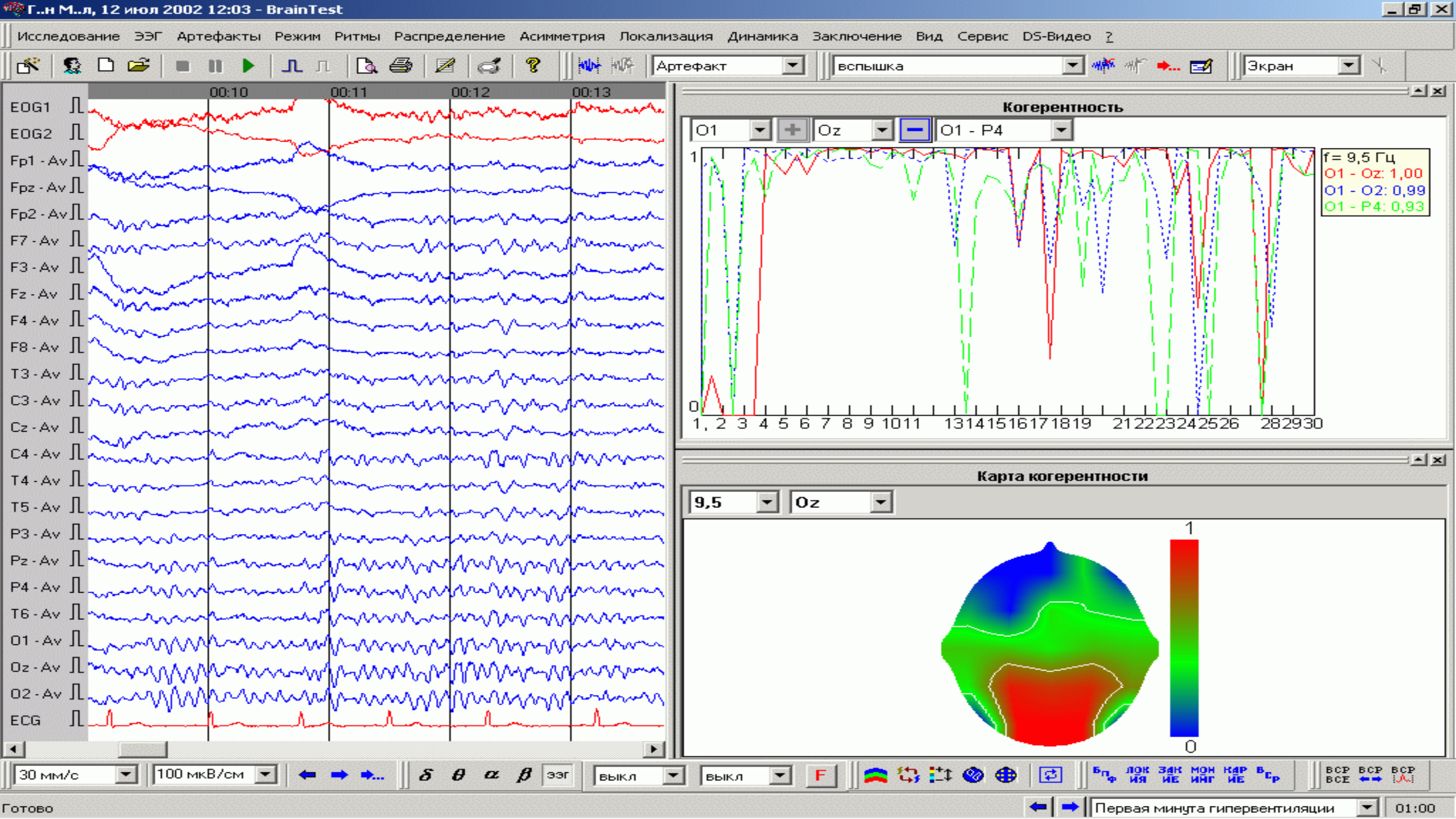
Gait and Balance Disorders

- Both are common in elderly
- Gait disorders can be due to lower extremity problems or neurological problems
- Balance problems may be caused by orthopedic dysfunction, low back problems,

  cerebellar dysfunction or inner ear problems

Diagnostic Tests in Neurology

- Lumbar puncture
  - Has been used for over 100 years
  - Tests CSF for infections, pressure, and lab data such as glucose, proteins and WBC
- EEG – electroencephalography
  - Measure sequential EEGs to look for change in brain function
  - Evoked potentials show brain activity
  - A new approach is brain mapping in color


Г..н М..л, 12 июл 2002 12:03 - BrainTest
Исследование ЭЭГ Артефакты Режим Ритмы Распределение Асимметрия Локализация Динамика Заключение Вид Сервис DS-Видео ?
Артефакт
вспышка
Экран
00:10
00:11
00:12
00:13
EOG1
EOG2
Fp1 - Av
Fpz - Av
Fp2 - Av
F7 - Av
F3 - Av
Fz - Av
F4 - Av
F8 - Av
T3 - Av
C3 - Av
Cz - Av
C4 - Av
T4 - Av
T5 - Av
P3 - Av
Pz - Av
P4 - Av
T6 - Av
O1 - Av
Oz - Av
O2 - Av
ECG
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1, 2 3 4 5 6 7 8 9 1011 13141516171819 21222324252628293 0
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Первая минута гипервентиляции
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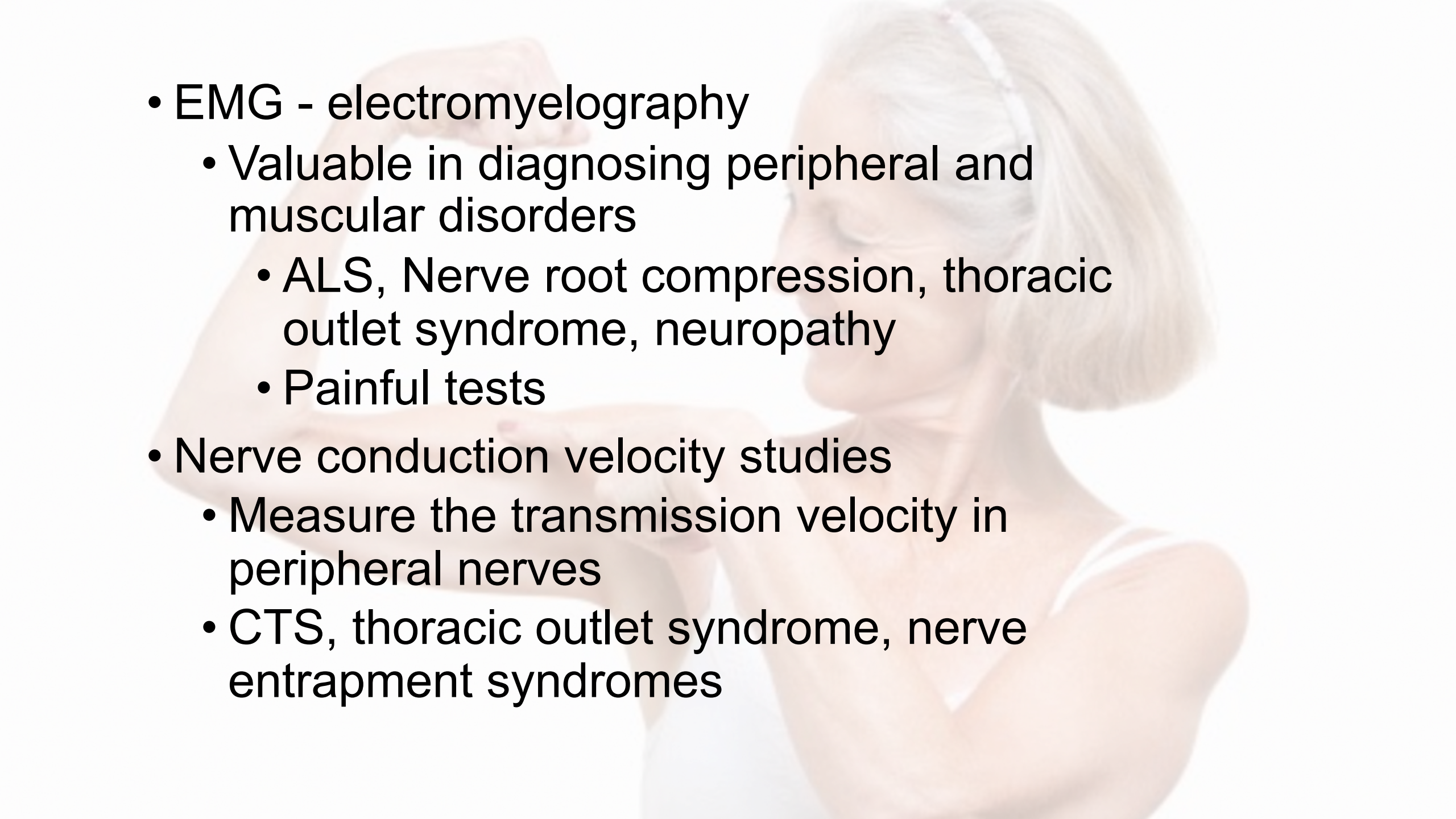
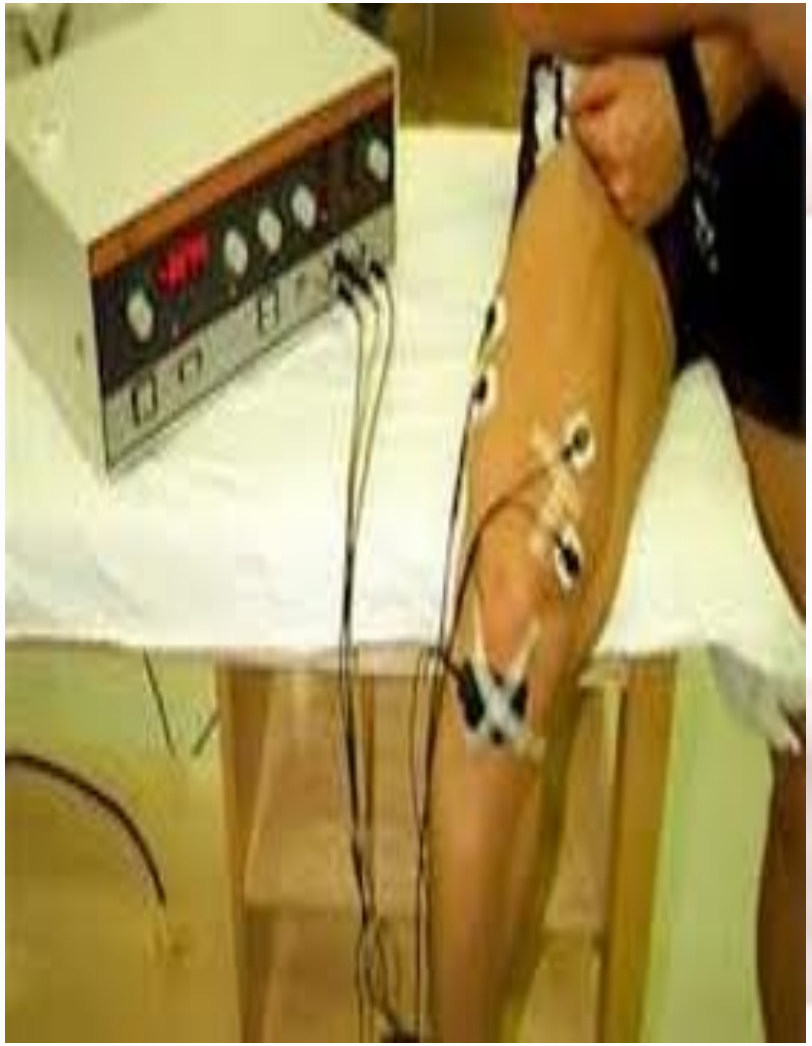


- EMG - electromyelography
  - Valuable in diagnosing peripheral and muscular disorders
    - ALS, Nerve root compression, thoracic outlet syndrome, neuropathy
    - Painful tests
- Nerve conduction velocity studies
  - Measure the transmission velocity in peripheral nerves
  - CTS, thoracic outlet syndrome, nerve entrapment syndromes

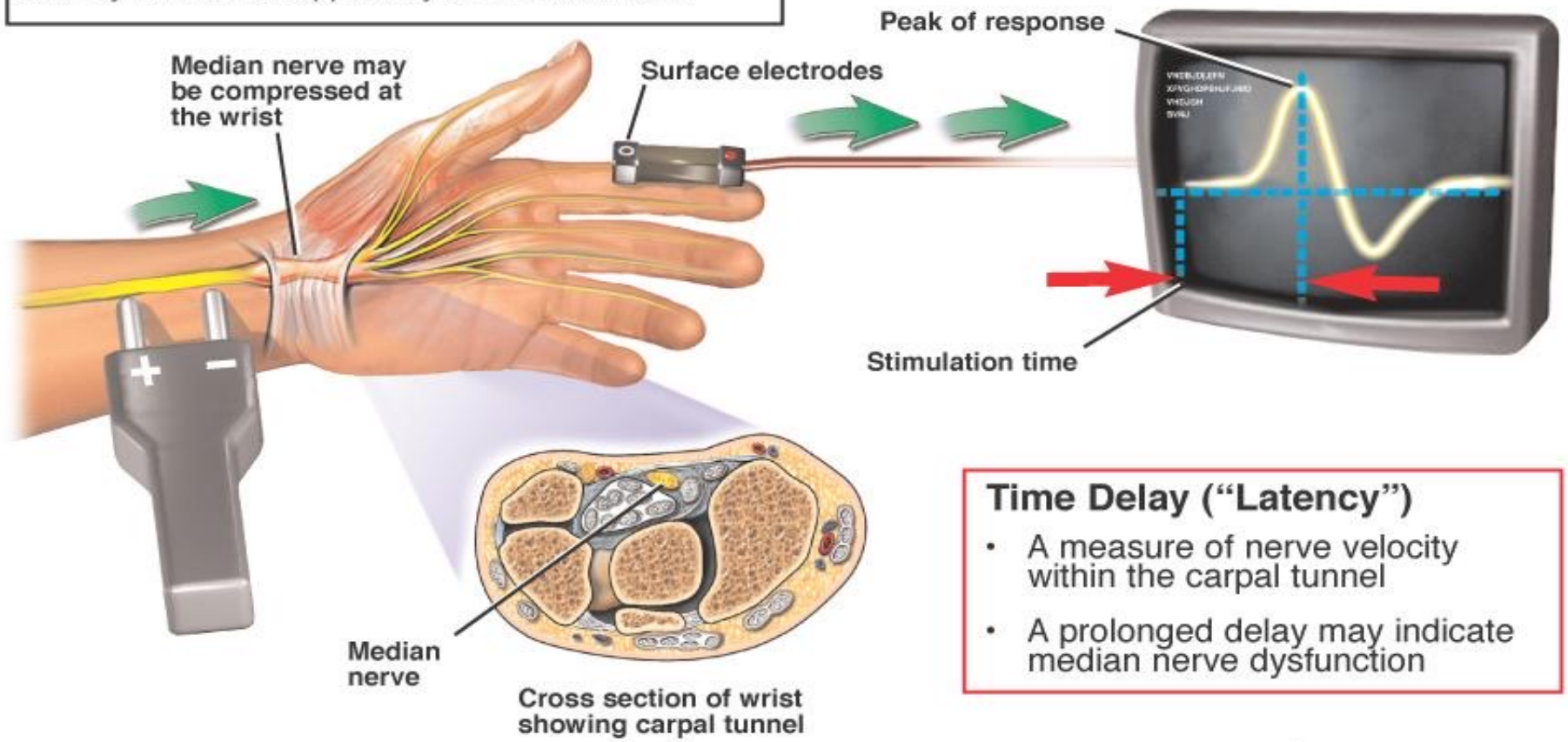


NCVS

Sensory Nerve Conduction Studies (Electrodiagnosis) of the Median Nerve Across the Carpal Tunnel

Surface stimulation of the median nerve at the wrist. This sends a nerve impulse through the carpal tunnel into the hand and activates the specific muscles and sensory structures supplied by the median nerve.

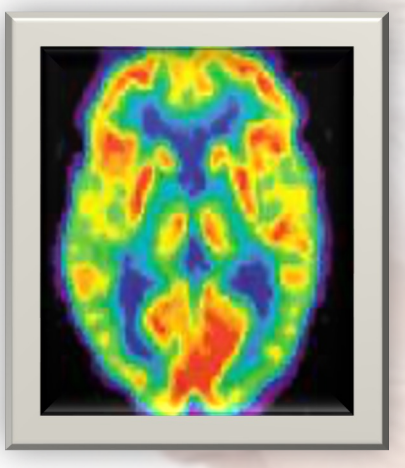
Recorded sensory nerve response at the index finger from the median nerve stimulation at the wrist



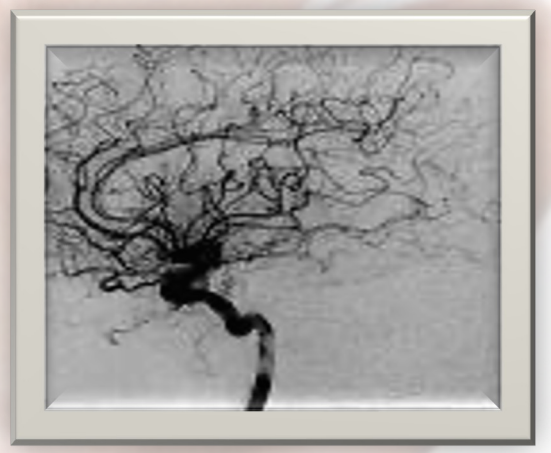
Time Delay ("Latency")

- A measure of nerve velocity within the carpal tunnel
- A prolonged delay may indicate median nerve dysfunction

- Neuroradiology
  - CT scans
  - MRI scans
  - PET scans
  - CT angiography
- Angiography
  - Injection of contrast dye
  - The gold standard of brain vascular diagnosis

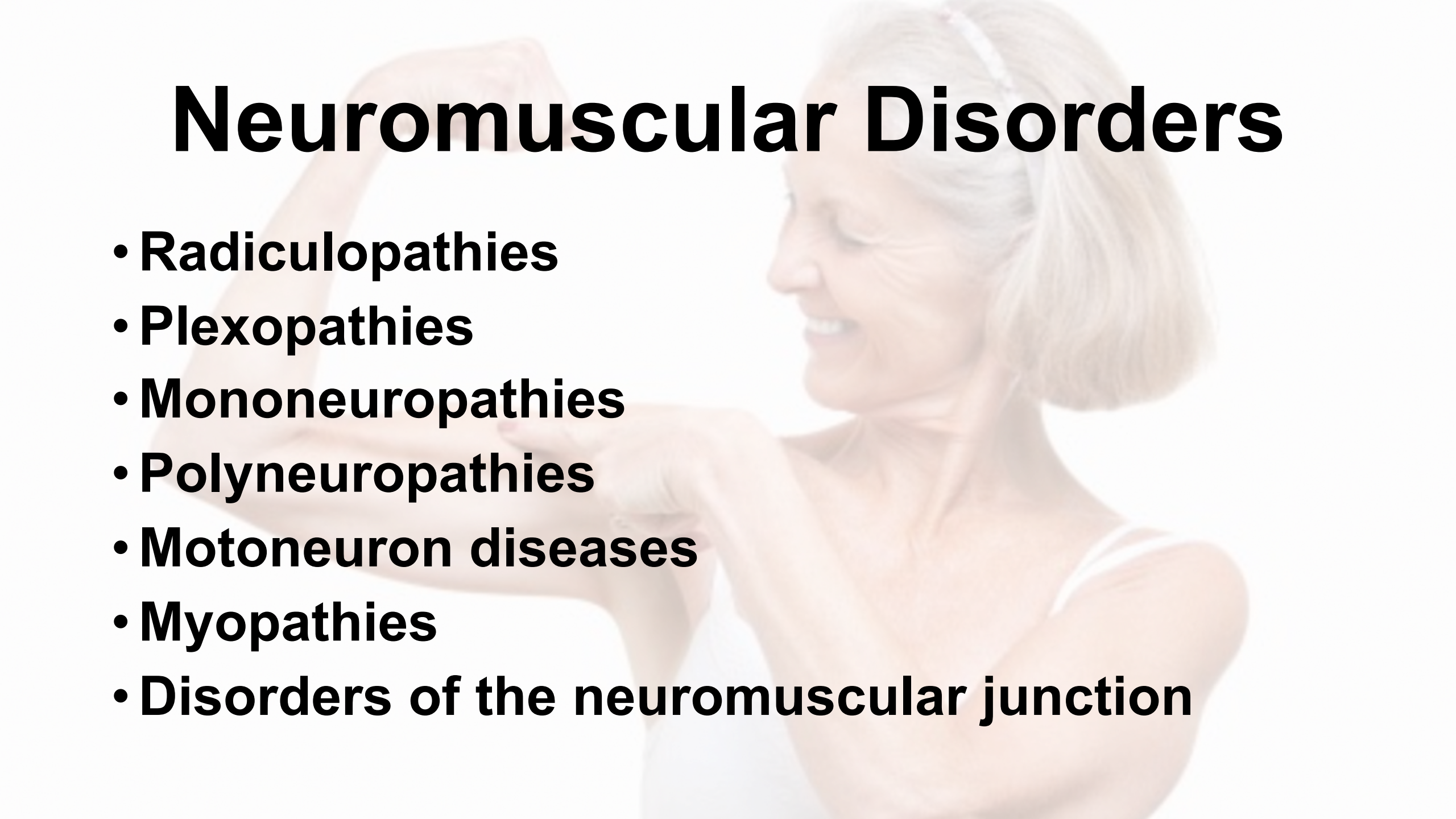


- Ruptured Brain Aneurysm



Ruptured Brain Aneurysm

Neuromuscular Disorders



- Radiculopathies
- Plexopathies
- Mononeuropathies
- Polyneuropathies
- Motoneuron diseases
- Myopathies
- Disorders of the neuromuscular junction

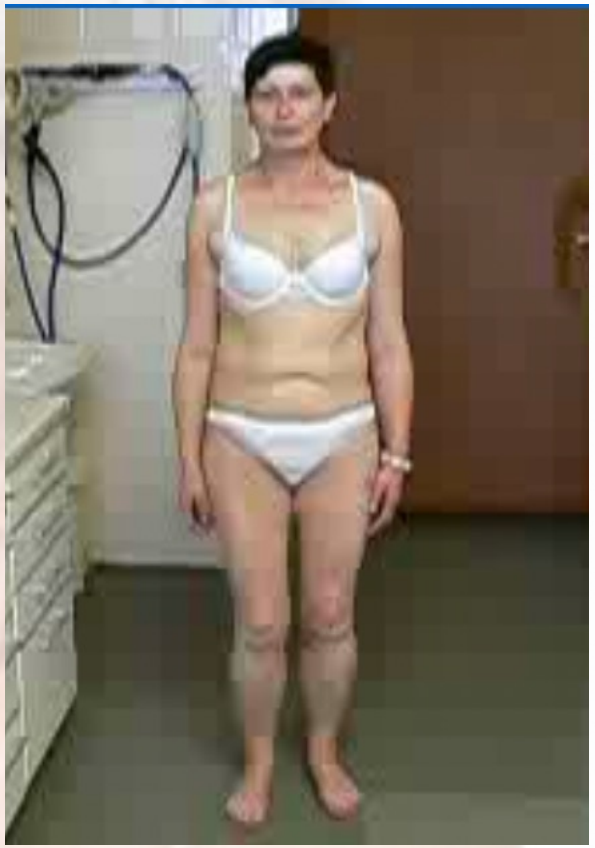
Symptoms of muscle disease

- Muscle fatigue, exercise intolerance in general
- Proximal and symmetric weakness
  - Hyperextension of the knee
  - Increased lordosis of the lumbar spine, scoliosis
  - Contractures, tight Achilles tendons
- Myopathic face
- Muscle atrophy
- Myotonia
  - Muscles are unable to relax after they contract
- Tendon reflexes are normal or decreased

Diagnosis of muscle diseases

- Creatinine kinase levels increased in many myopathies (sign of muscle necrosis)
- EMG / ENG - differentiation between neurogenic and myogenic weakness
- Muscle biopsy - signs of muscle fiber abnormality, and inflammation
- Genetic testing

Is it really myopathy?

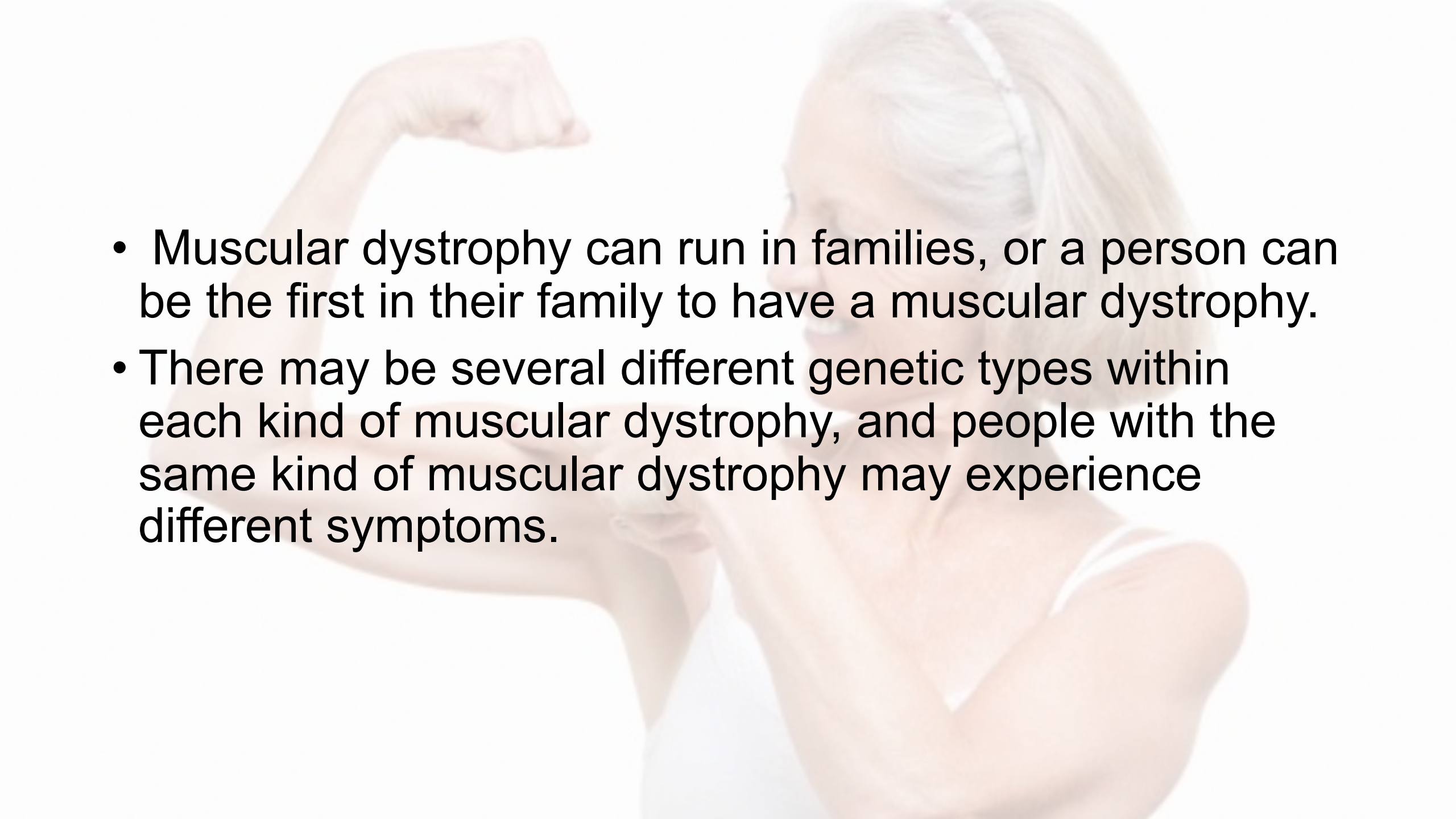


Types of muscle diseases

- Hereditary muscle diseases
  - Muscle dystrophies
  - Muscle channelopathies
  - Mitochondrial myopathies
  - Metabolic myopathies
- Acquired muscle diseases
  - Inflammatory myopathies
  - Endocrine and toxic myopathies
  - Infectious muscle diseases

Muscle Dystrophies

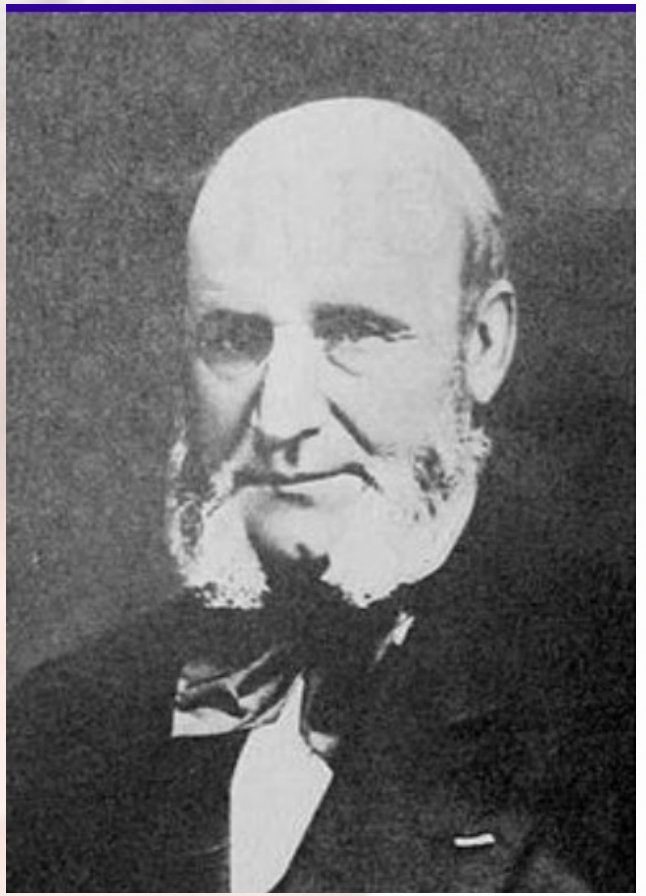
- Muscular dystrophies are a group of muscle diseases caused by mutations in a person's genes.
- Over time, muscle weakness decreases mobility, making everyday tasks difficult.
- There are many kinds of muscular dystrophy, each affecting specific muscle groups, with signs and symptoms appearing at different ages, and varying in severity.

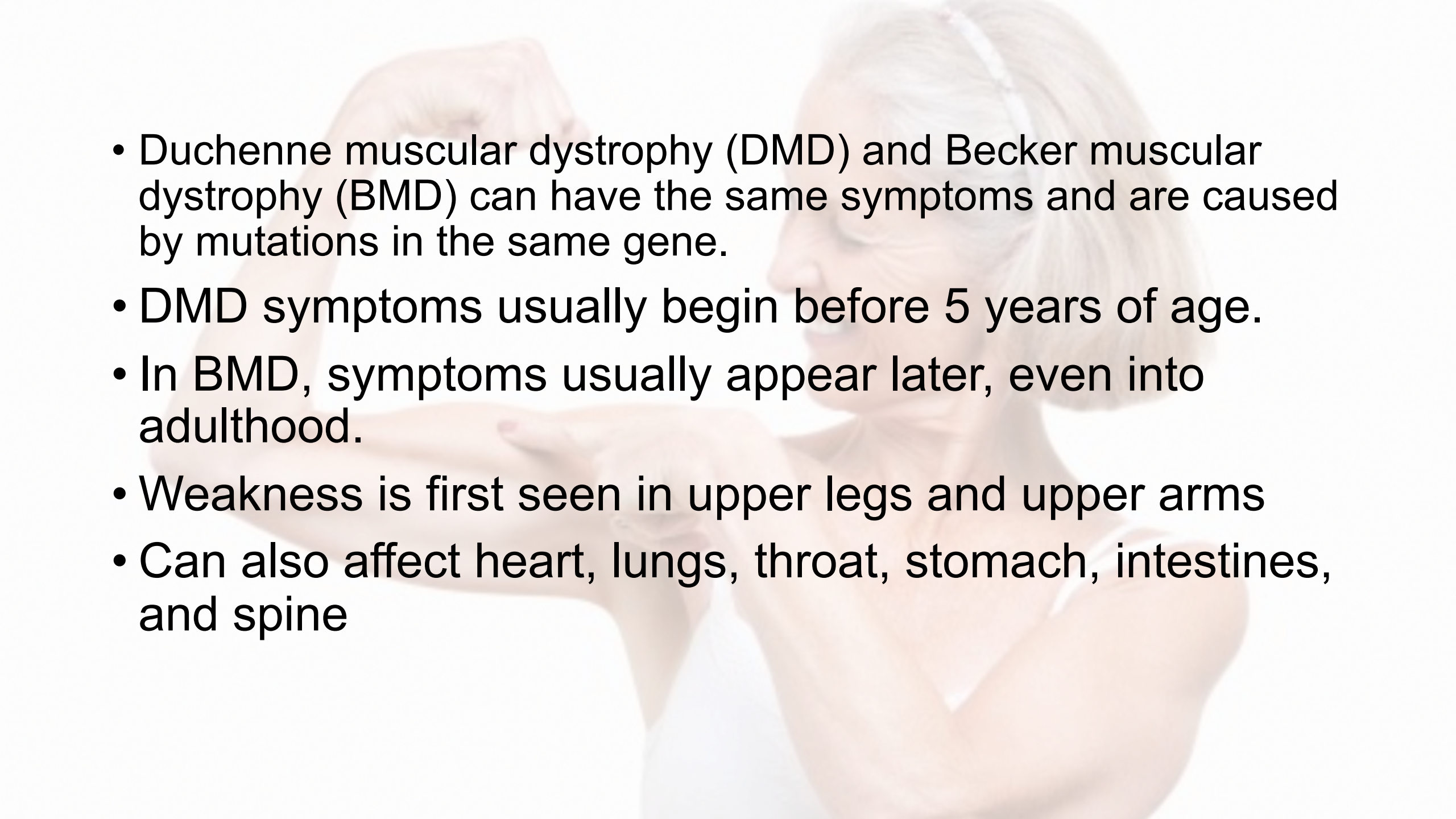


- Muscular dystrophy can run in families, or a person can be the first in their family to have a muscular dystrophy.
- There may be several different genetic types within each kind of muscular dystrophy, and people with the same kind of muscular dystrophy may experience different symptoms.

Duchenne Muscular Dystrophy (DMD)

- Dystrophinopathy
  - First described in 1881 - Dystrophin gene discovered in the early 1980s
- Cause - deficiency of dystrophin, resulting in progressive loss of muscle fibers
  - Becker's type - reduced amount of dystrophin with a more benign course
- X chromosome linked
  - 14 in 100,000 births - occurs in boys and girls are carriers



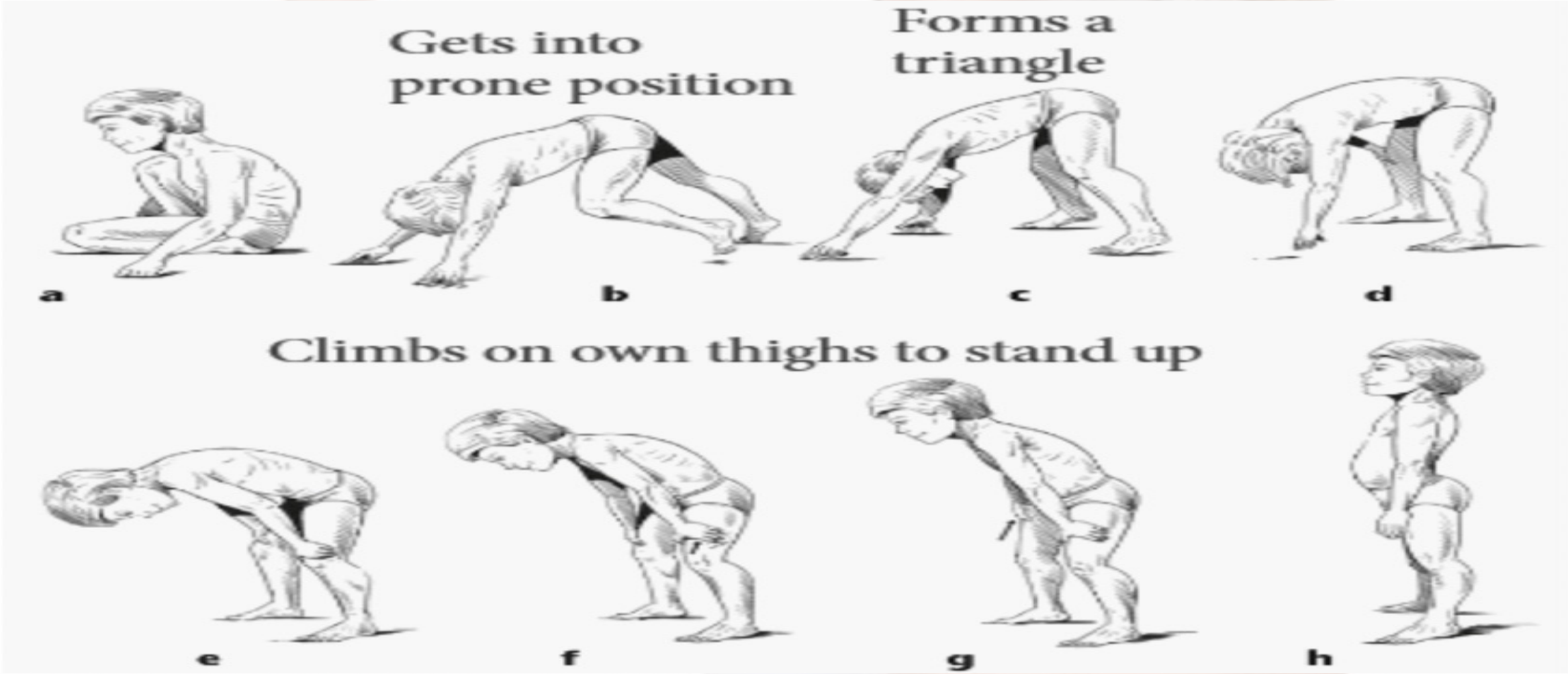


- Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) can have the same symptoms and are caused by mutations in the same gene.
- DMD symptoms usually begin before 5 years of age.
- In BMD, symptoms usually appear later, even into adulthood.
- Weakness is first seen in upper legs and upper arms
- Can also affect heart, lungs, throat, stomach, intestines, and spine

Duchenne Muscular Dystrophy (DMD)

- Onset at 3 to 5 years old
- Initial symptoms include difficulty getting up from deep positions and climbing steps with a waddling gait
- Weakness is more pronounced in limb-girdle muscles and trunk erectors. The cranial muscles are spared.
- Skeletal deformities
- Inability to walk by 9-11 years old
- Death usually occurs by the 3rd decade from respiratory insufficiency and cardiomyopathy

Grower's Sign

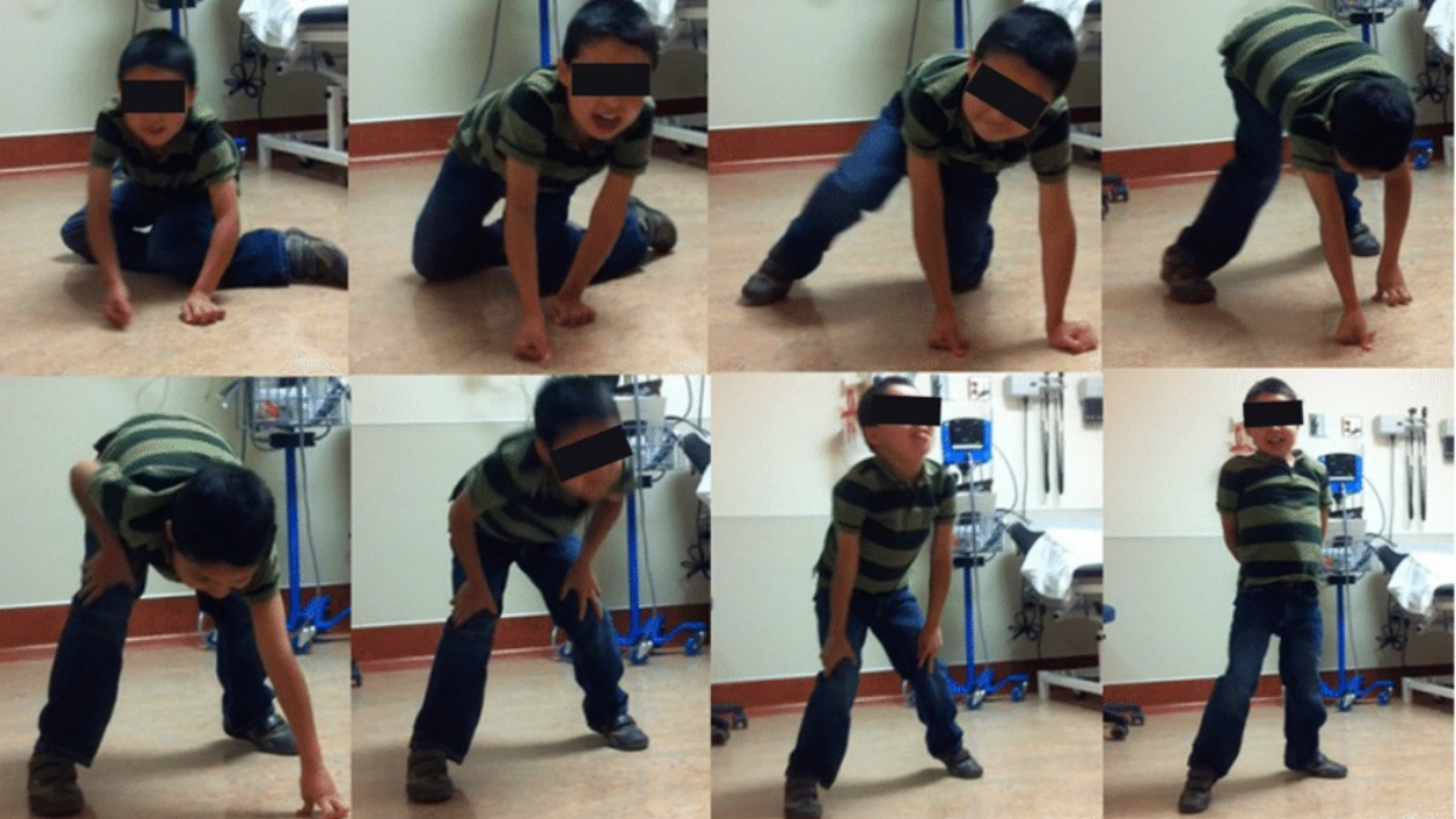


Gowers' Sign

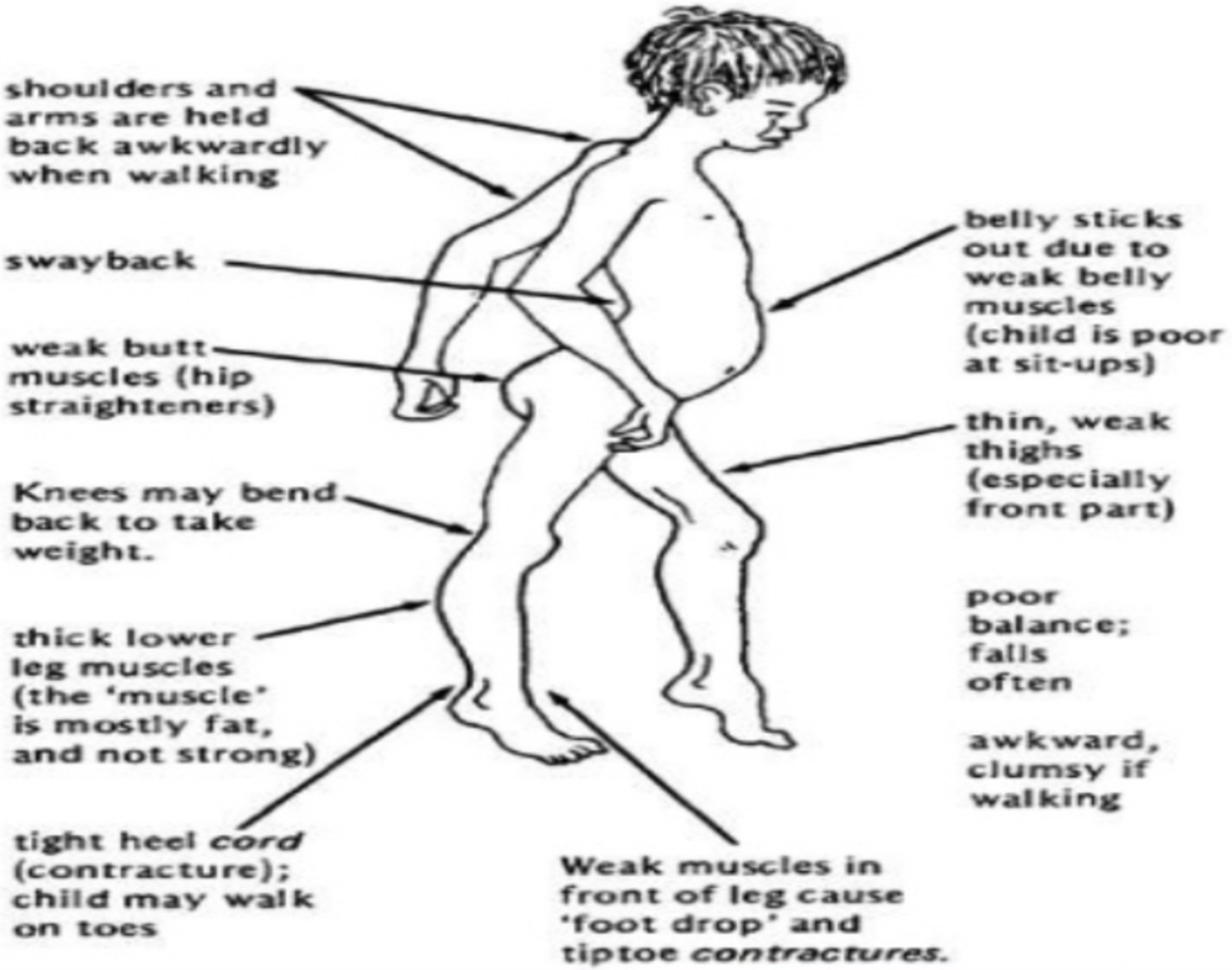
Duchenne Muscular Dystrophy (DMD)

DMD is a genetically transmitted condition that has an X - linked recessive mode of inheritance.

Healthy carrier mothers transmit the condition to their male children.



DMD: Clinical manifestation

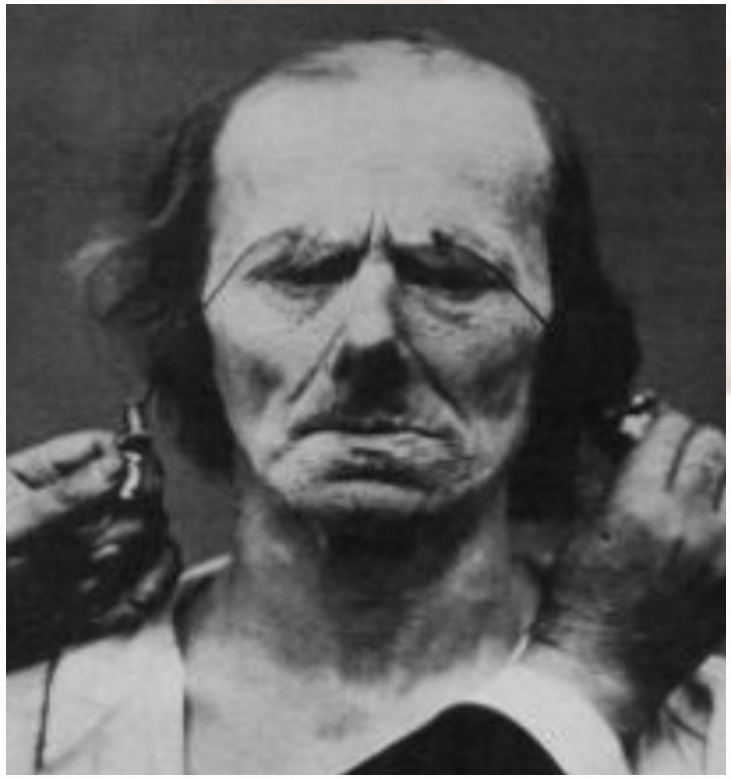


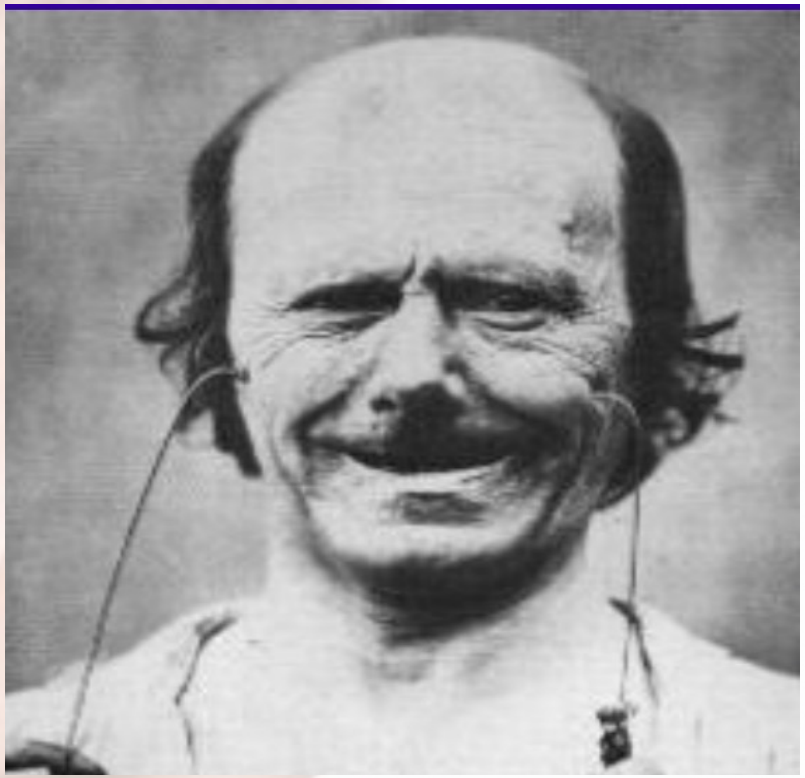
- **Onset : age 3–6 years**
- **Progressive weakness**
- **Pseudohypertrophy of calf muscles**
- **Spinal deformity**
- **Cardiopulmonary involvement**

Mild – moderate MR

Completely irrelevant side note

- Duchenne investigated facial expressions is a crude but effective experiment.
- He ”shocked the facial muscles using galvanic current” and tried to define ”facial expressions”



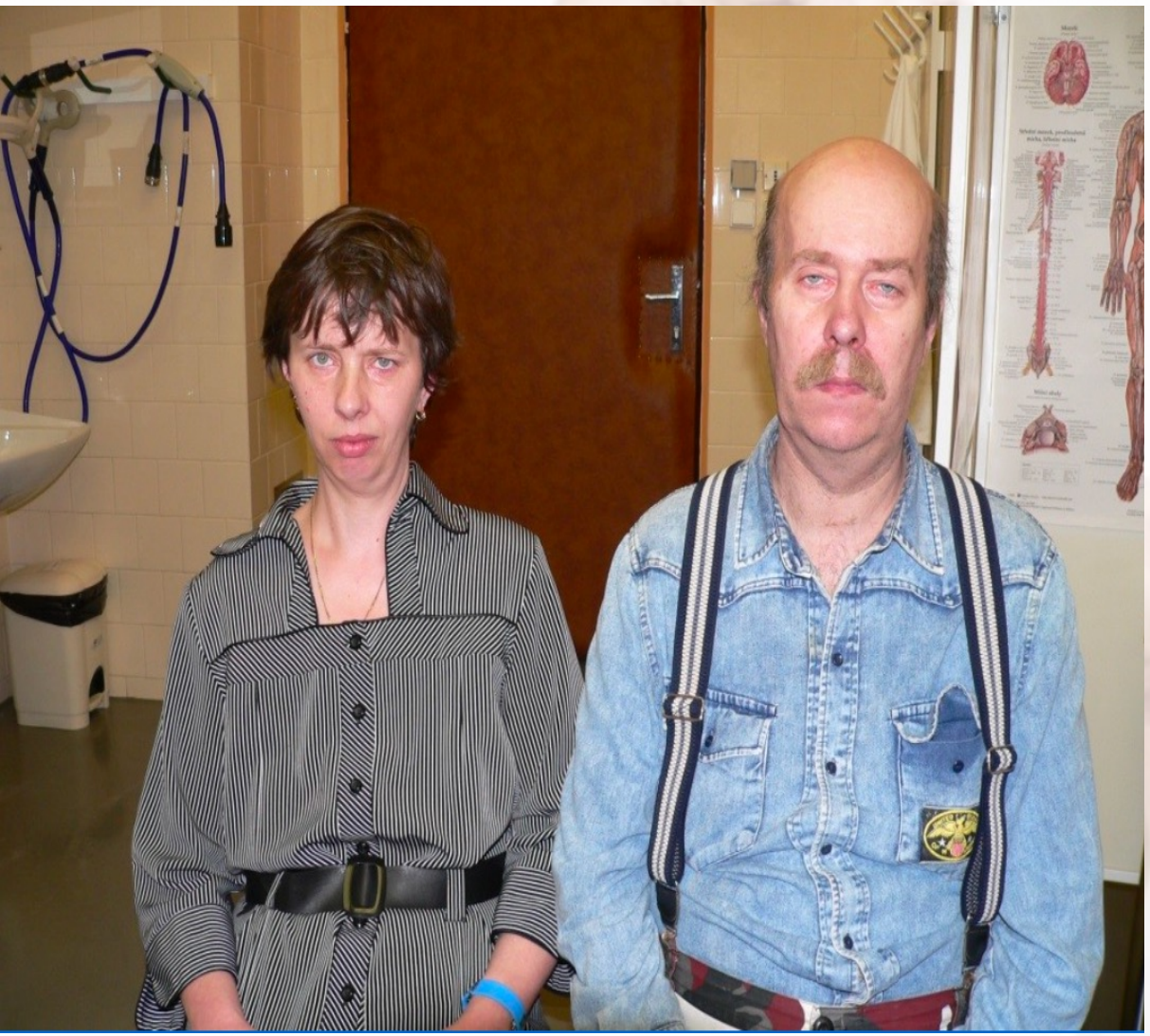


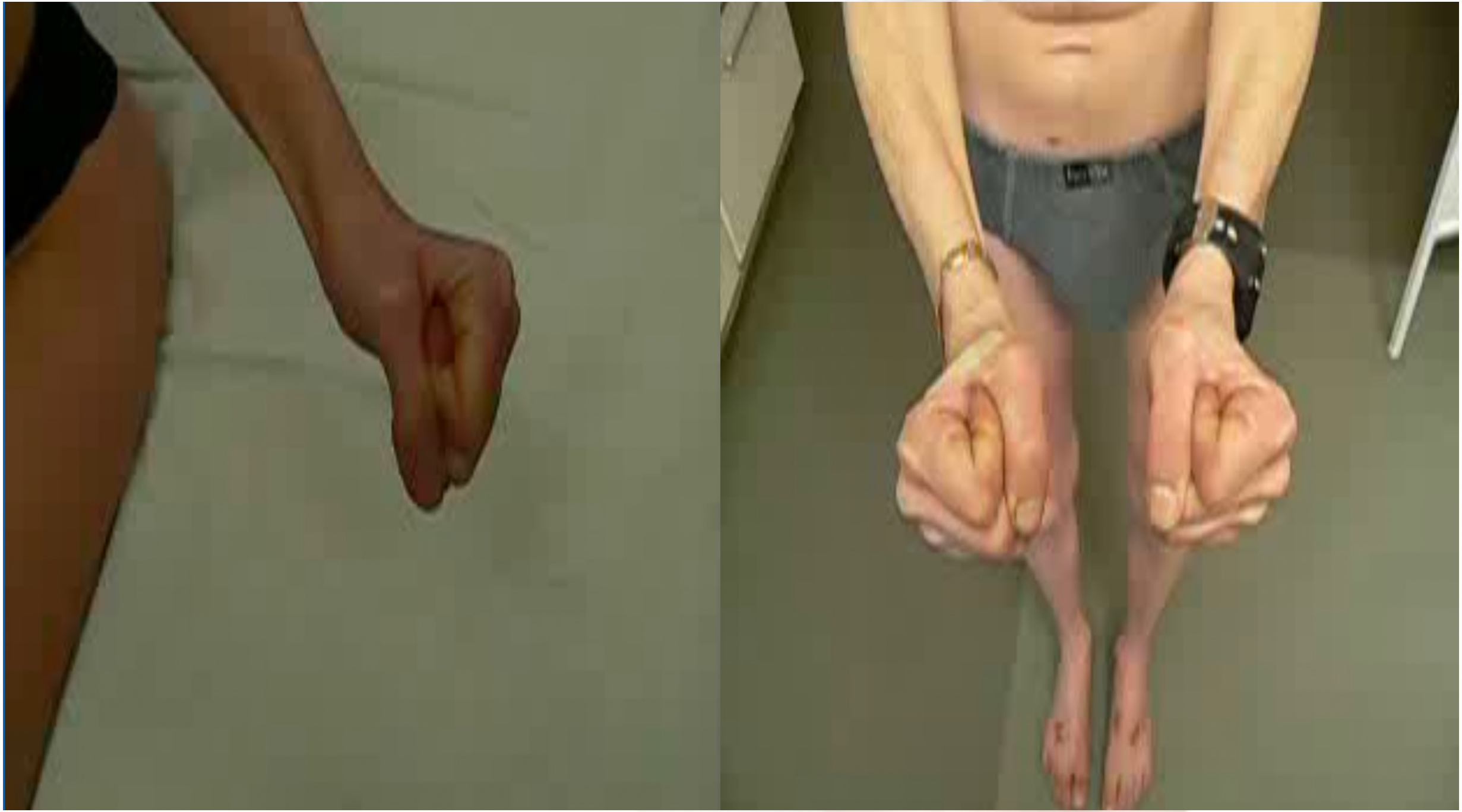
Myotonic Dystrophy

- About 8 in 100,000 people of all ages are affected
- Males and females equally
- Muscle weakness usually starts between 10–30 years of age, but ranges from birth to 70 years old.
- Which parts of the body show weakness first? Face, neck, arms, hands, hips, and lower legs
- What other parts of the body can be affected? Heart, lungs, stomach, intestines, brain, eyes, and hormone-producing organs

Myotonic Dystrophy
Is a multisystemic disease

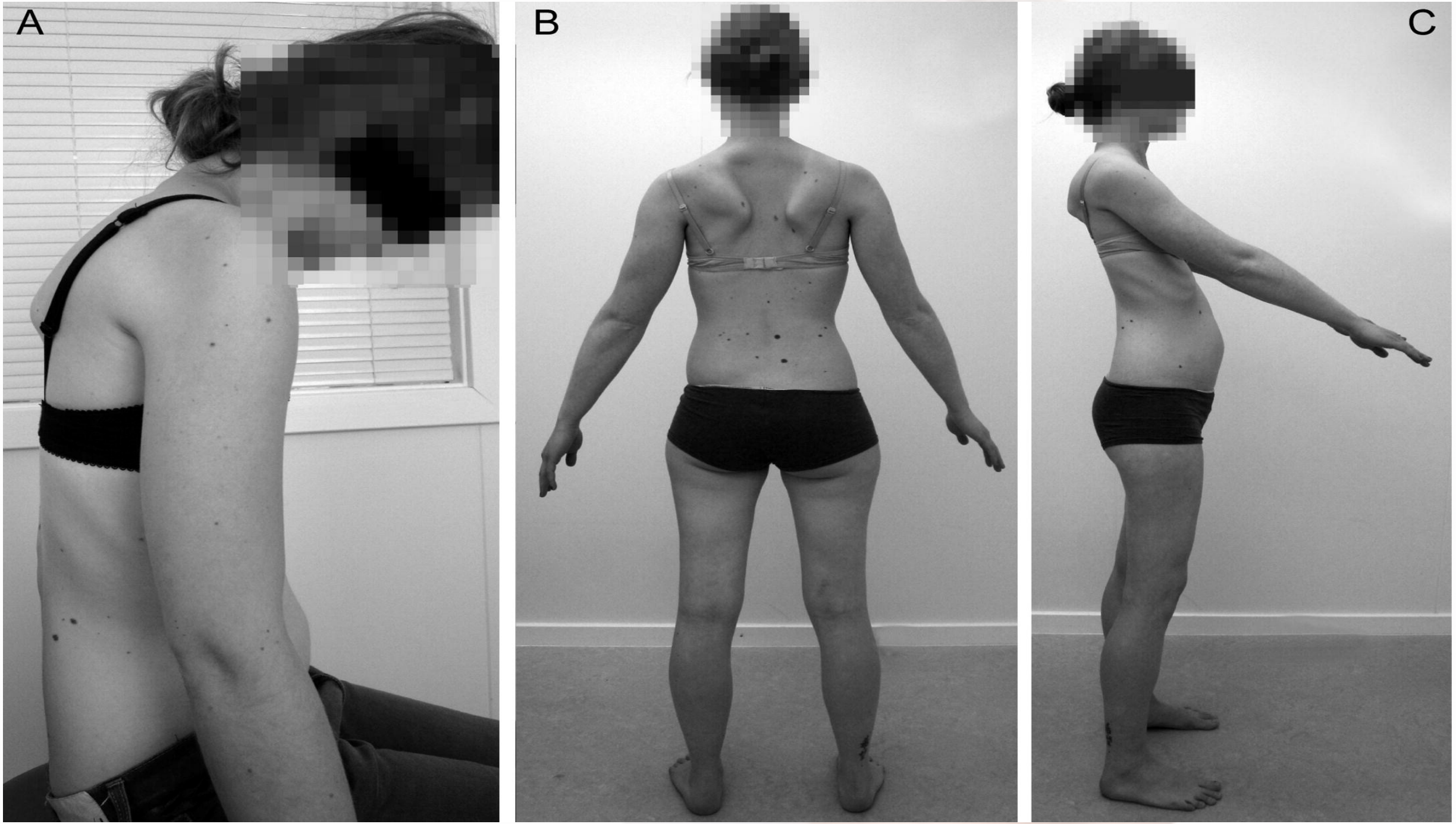
- Myotonia - hyperexcitability of muscle membrane which causes an inability of quick muscle relaxation
- Progressive muscular weakness and wasting, most prominent in cranial and distal muscles
- Cataracts, frontal balding, testicular atrophy
- Cardiac abnormalities, mental retardation





Limb-girdle dystrophies

- Limb-girdle muscular dystrophies are a large group of genetic diseases in which there is muscle weakness and wasting (muscular dystrophy).
- In most cases, both parents must pass on the non-working (defective) gene for a child to have the disease (autosomal recessive inheritance).
- About 2 in 100,000 people of all ages
- Males and females equally
- Muscle weakness can be in childhood or adulthood
- Body show weakness first in the upper arms, upper legs
- Can also affect heart, spine, hips, calves, and trunk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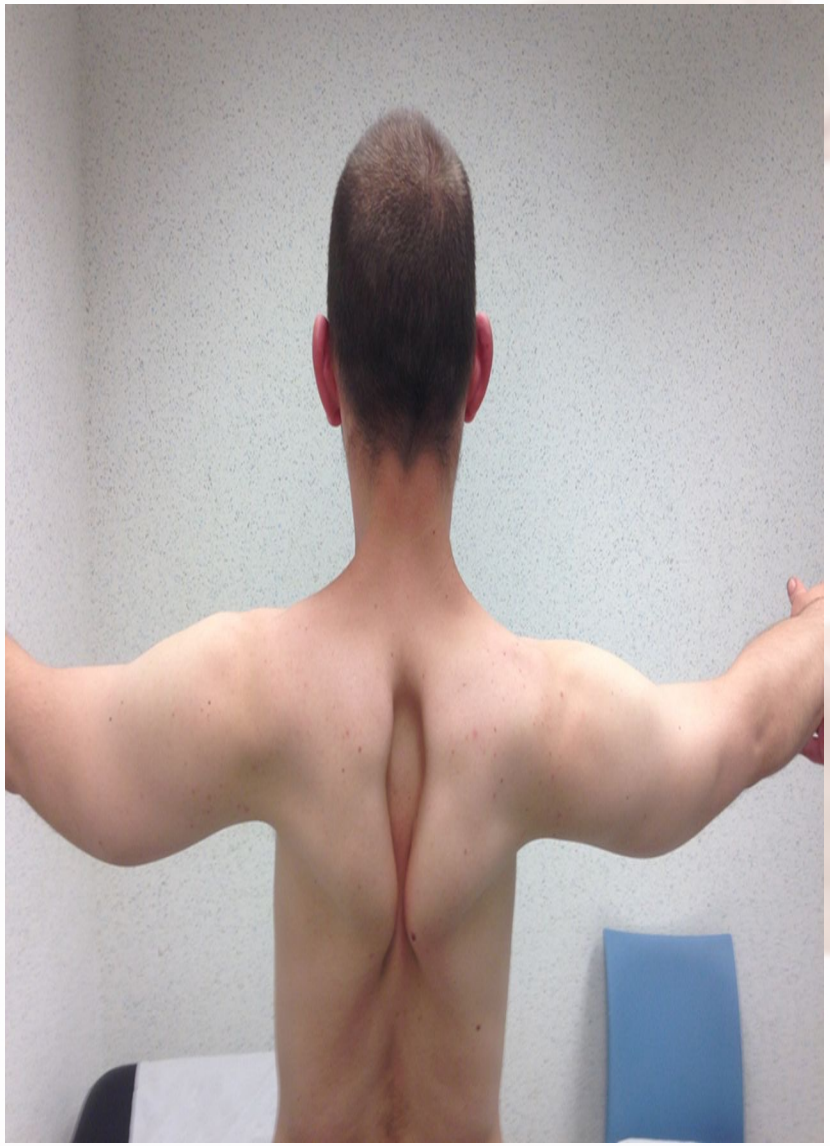

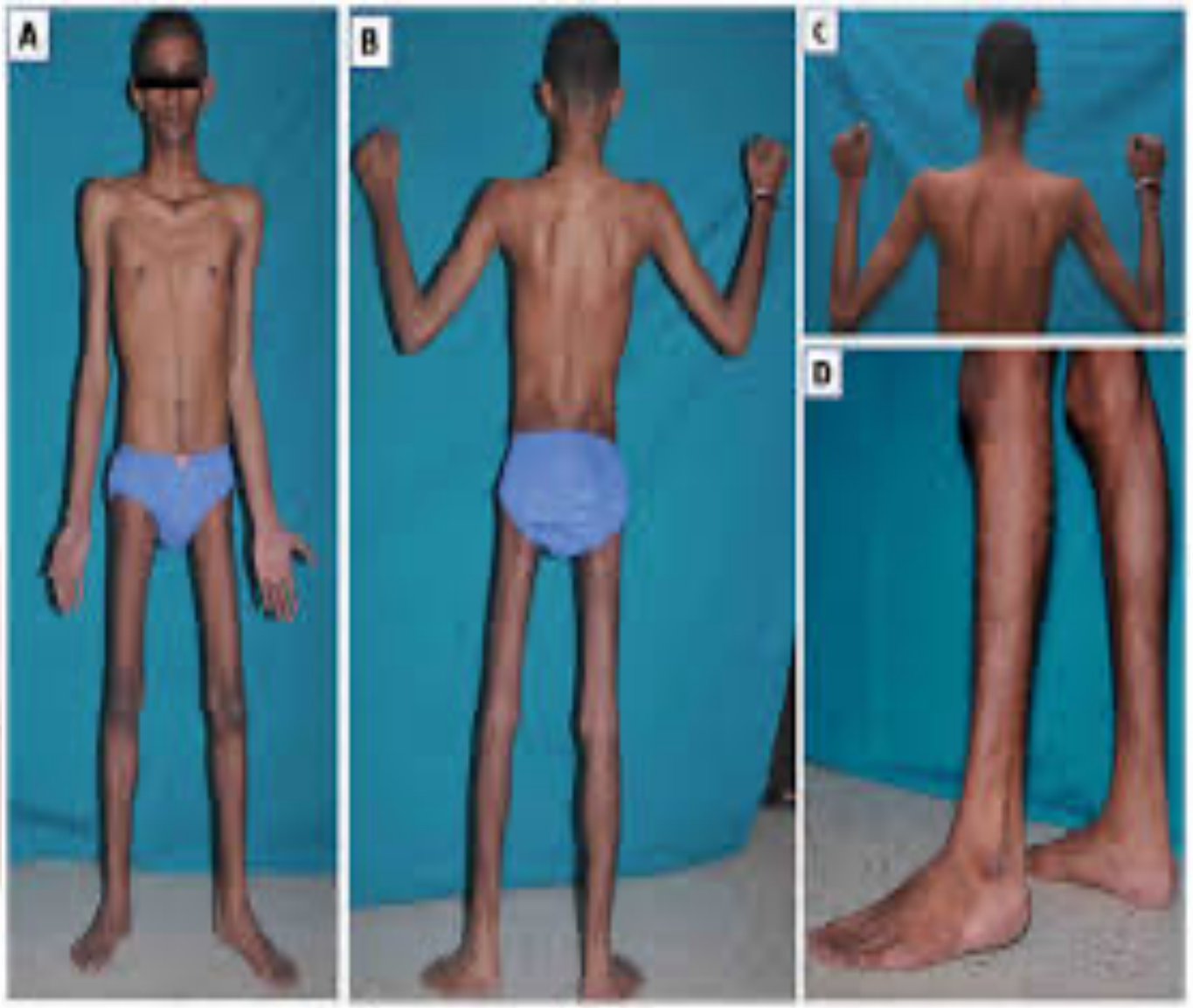
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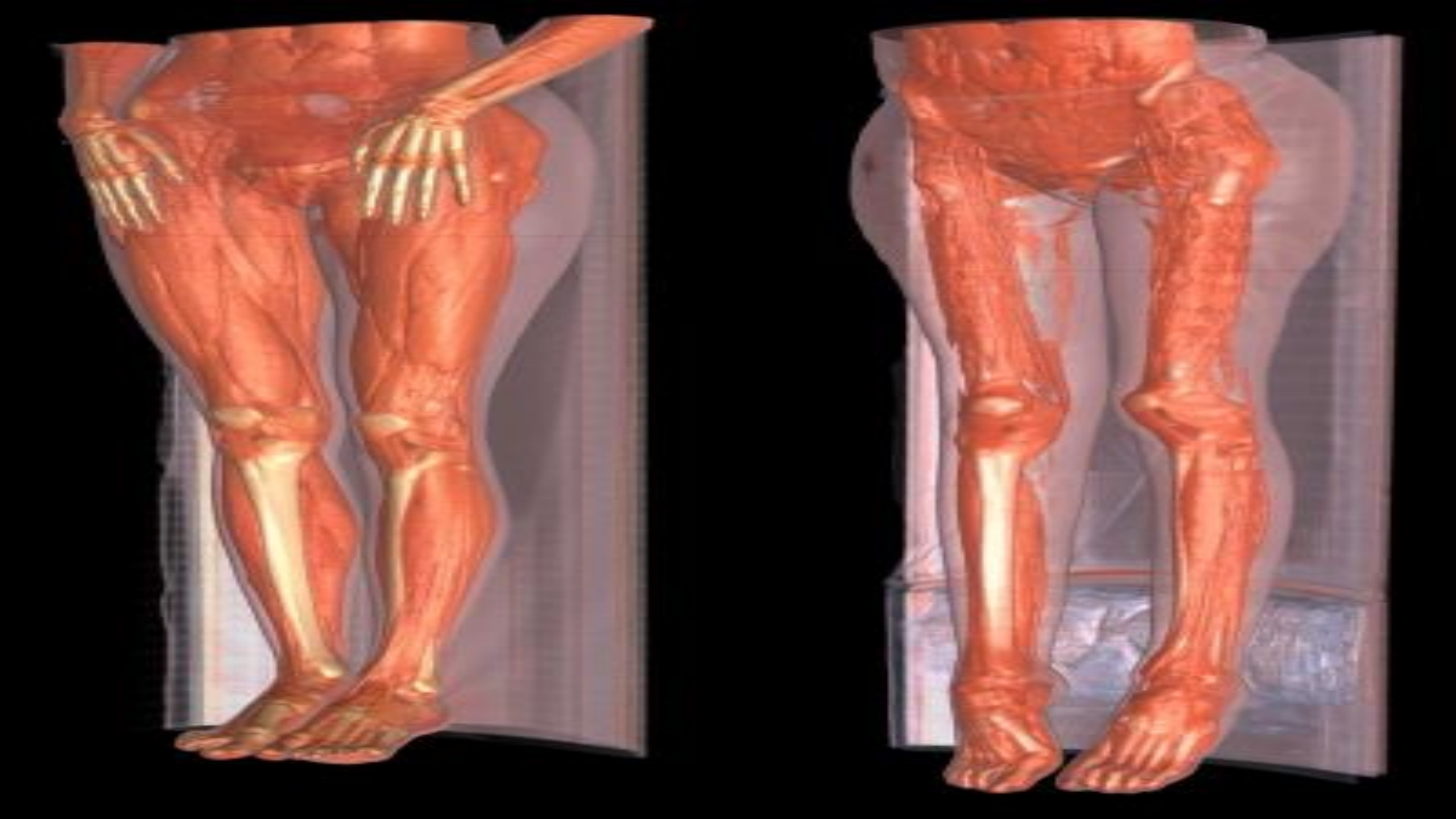
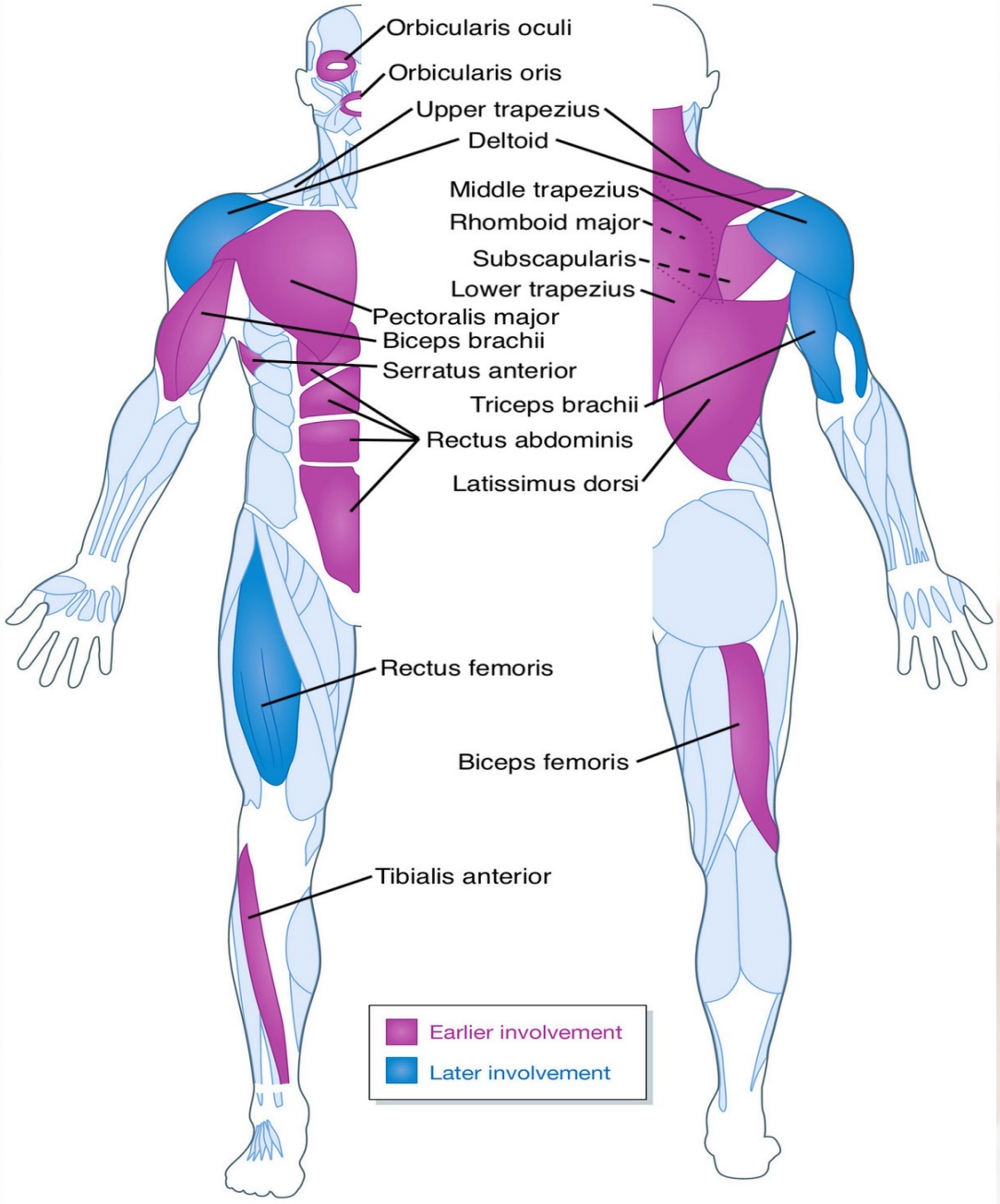
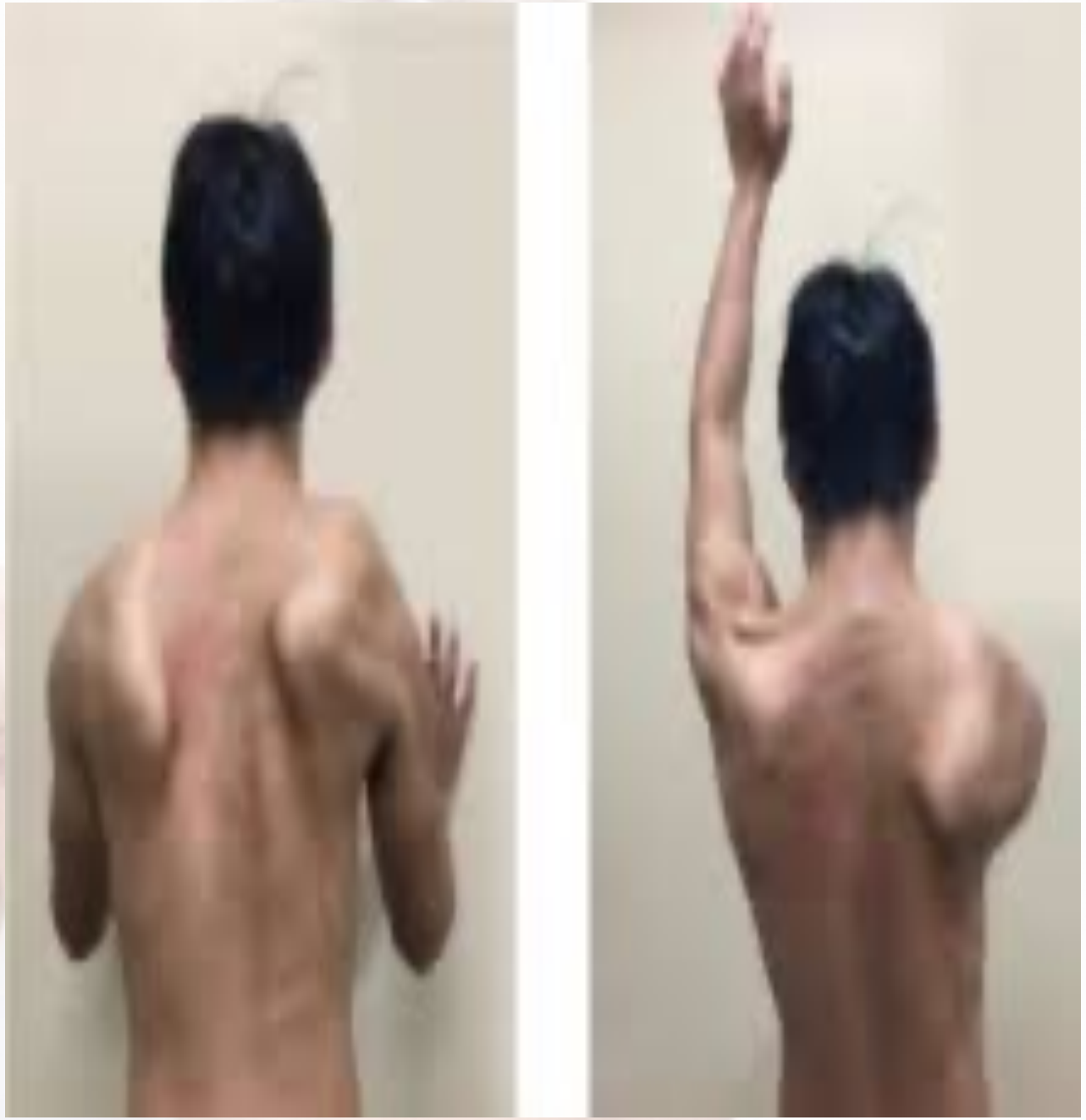
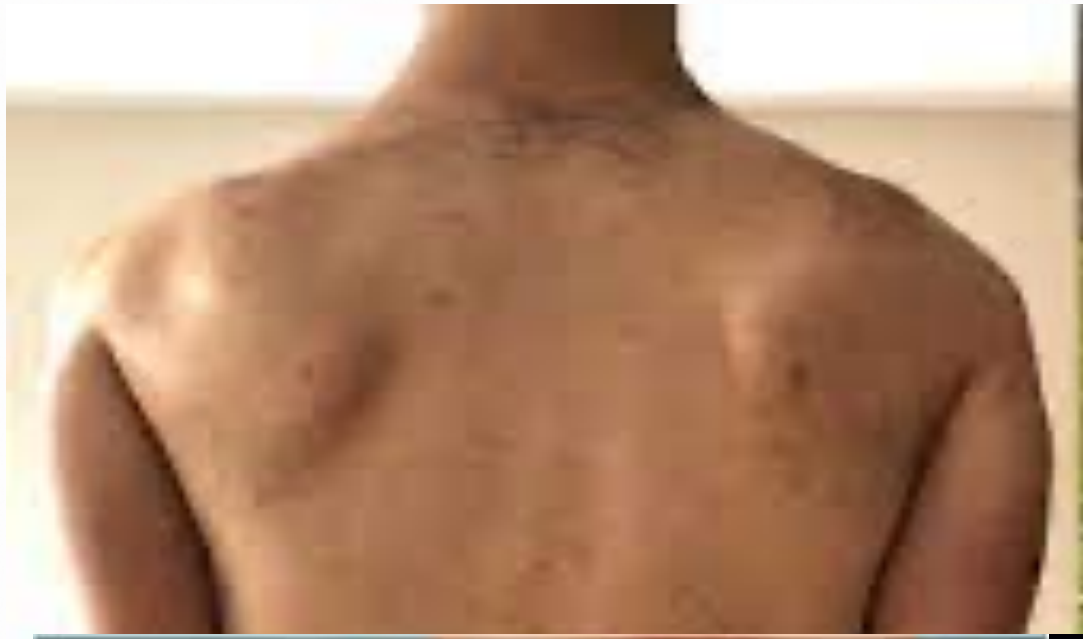


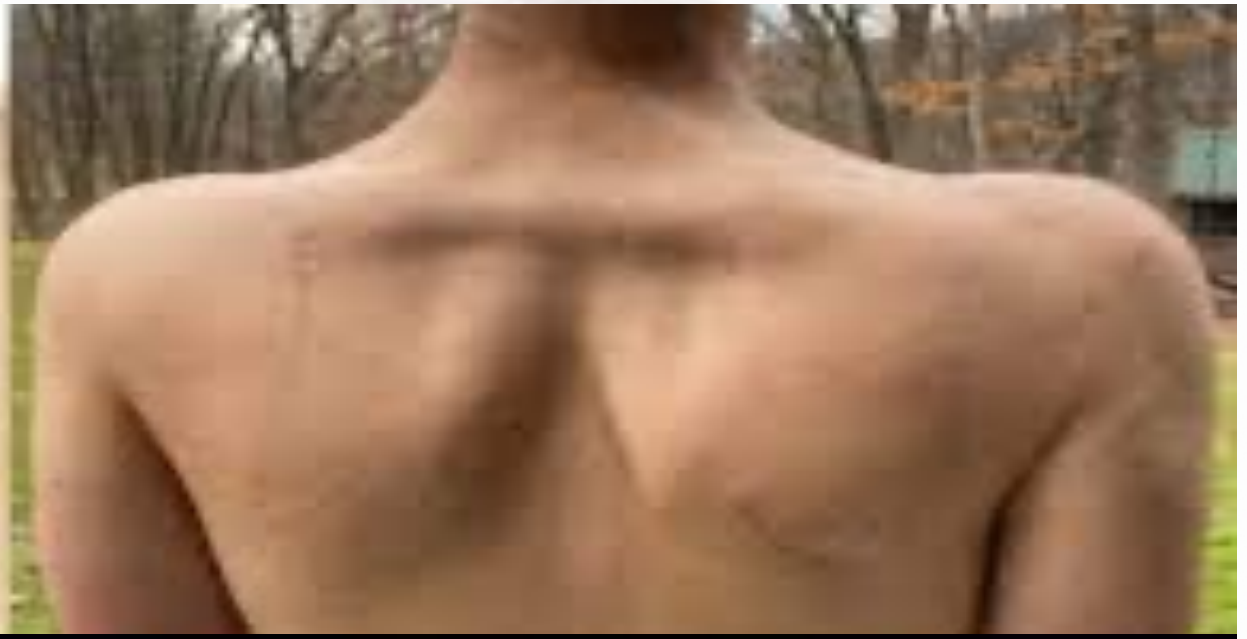
Facioscapulohumeral dystrophy
FSHD

- Prevalence: 1 in 20,000
- Age of onset: infancy to middle age
- Prevalence: 1 in 20,000
  involving the face, scapular, proximal arm and
- Progressive muscular weakness and atrophy involving the face, scapular and proximal arm and peroneal muscles
- Myopathic face, winging of the scapula, inability to raise the arms, foot drop
- Life span is not significantly affected


Orbicularis oculi
Orbicularis oris
Upper trapezius
Deltoid
Middle trapezius
Rhomboid major
Subscapularis
Lower trapezius
Pectoralis major
Biceps brachii
Serratus anterior
Triceps brachii
Rectus abdominis
Latissimus dorsi
Rectus femoris
Biceps femoris
Tibialis anterior
Earlier involvement
Later involvement



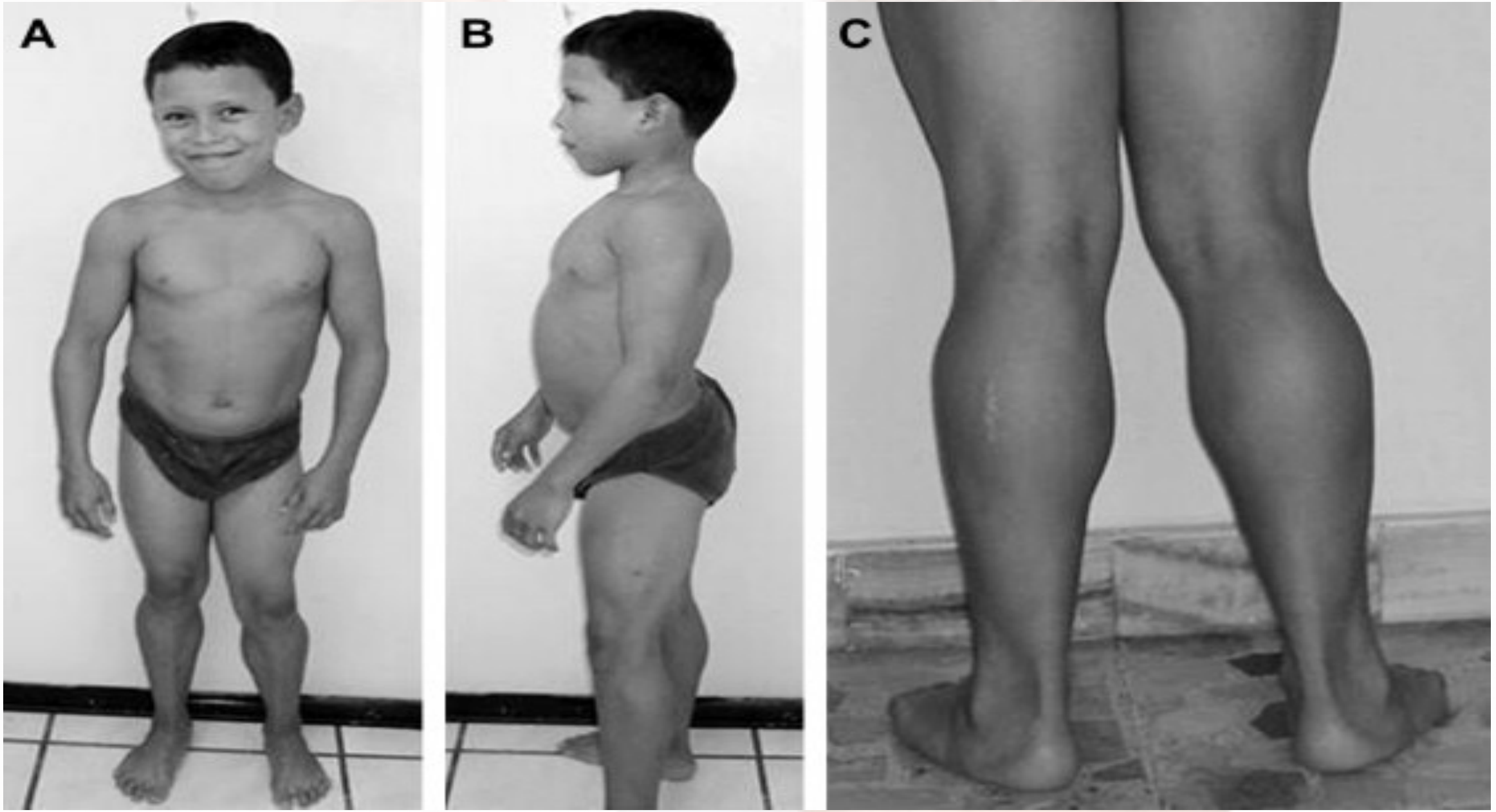




FSHD

Myotonia Congenita

- Myotonia congenita is an inherited neuromuscular disorder characterized by the inability of muscles to quickly relax after a voluntary contraction.
- The condition is present from early childhood, but symptoms can be mild.
- This condition can lead to muscle stiffness that affects your ability to move.
- Myotonia congenita can also increase the size of muscles and cause muscle cramping.
- Myotonia congenita affects the chloride channels on your muscle membrane. (Muscle Channelopathies)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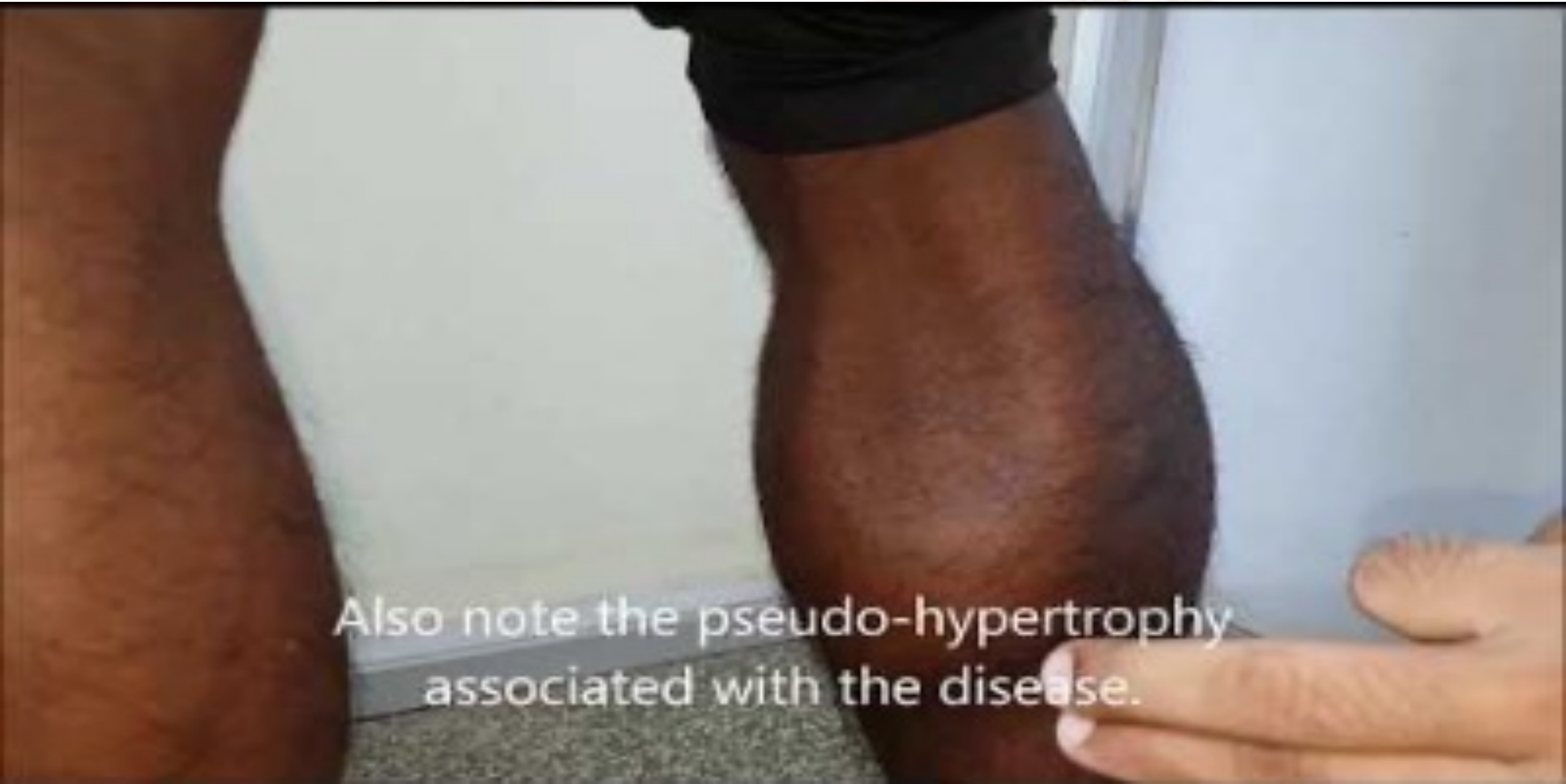
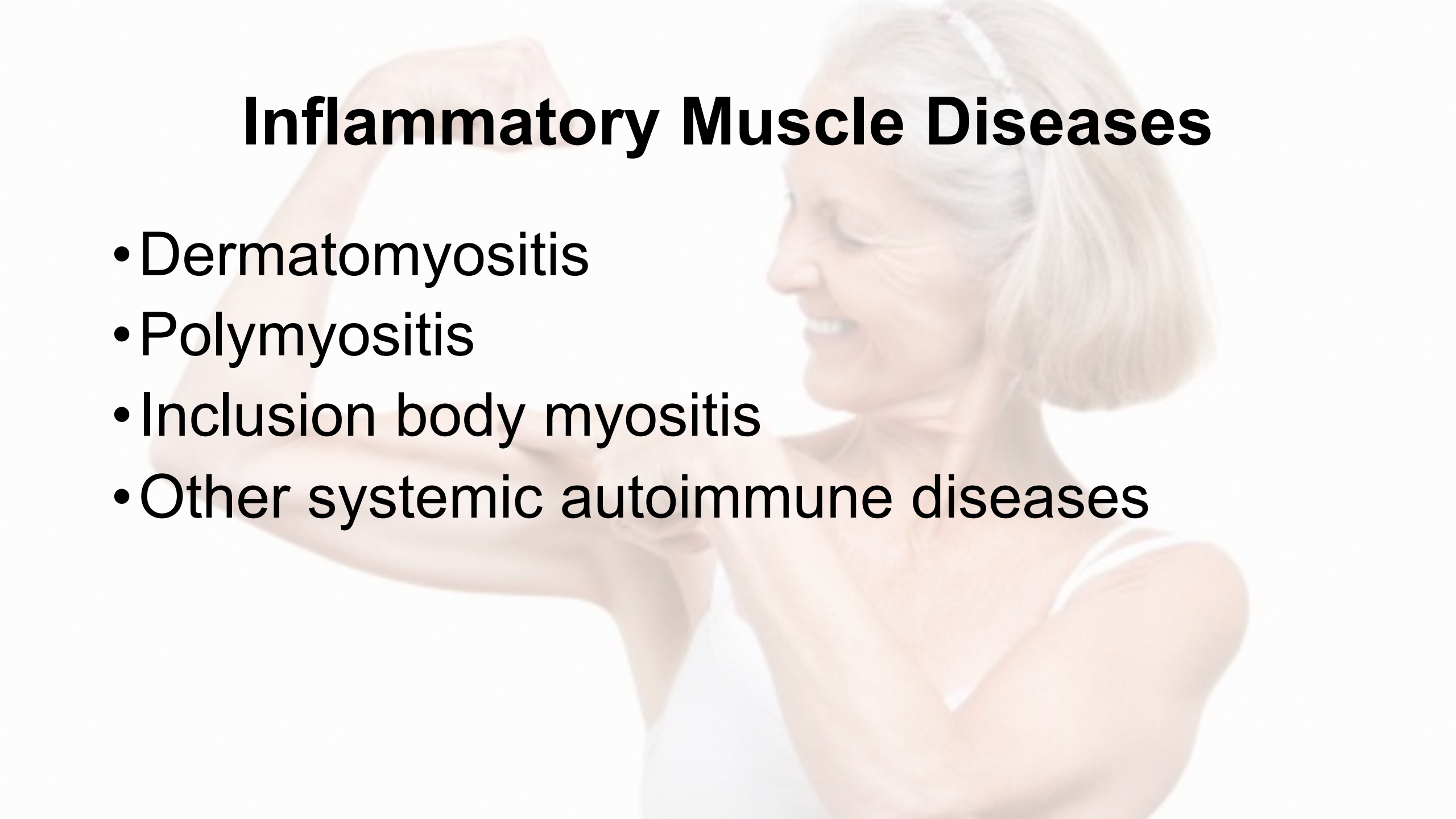
Myotonia Congenita



Inflammatory Muscle Diseases



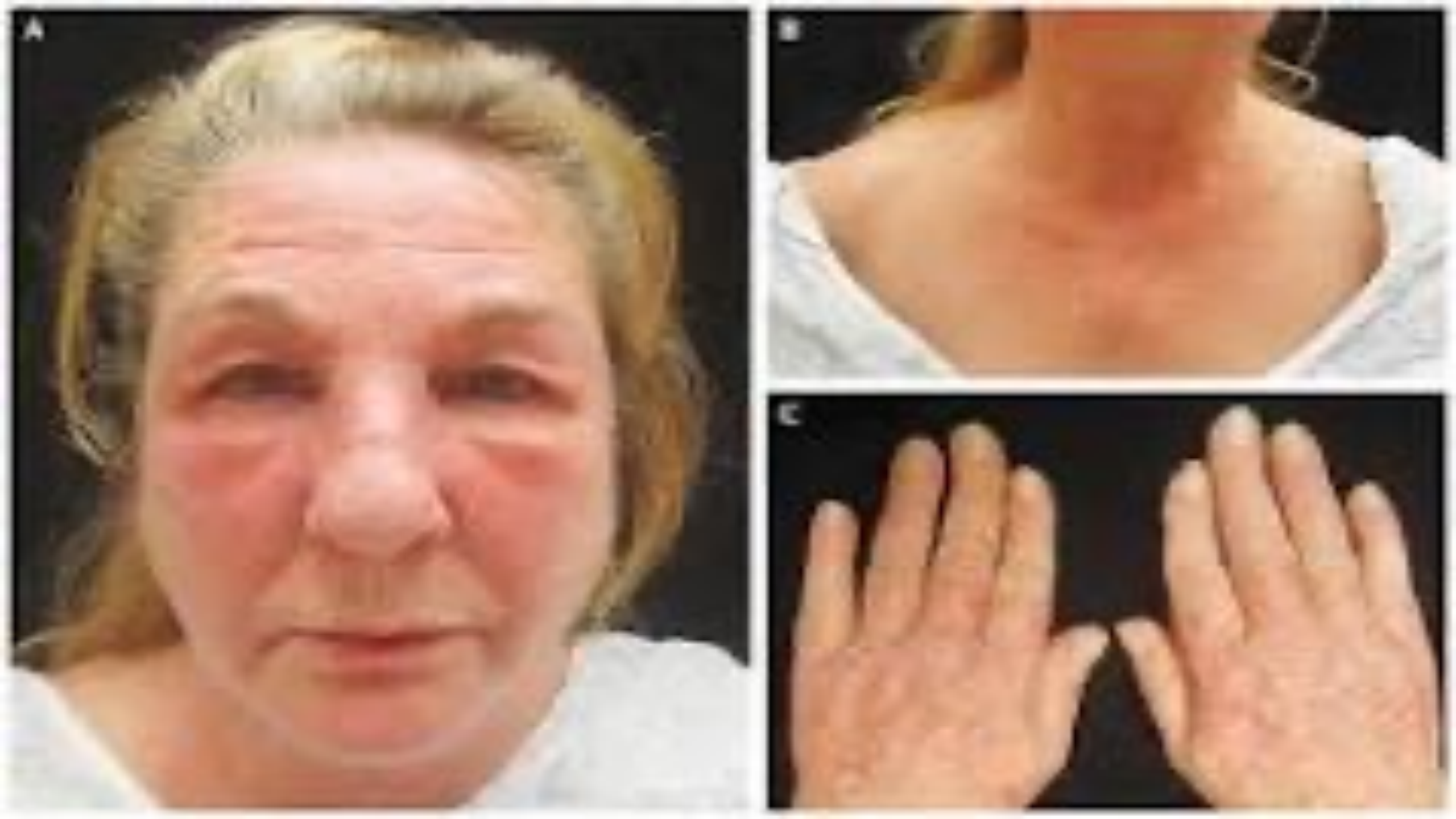
- Dermatomyositis
- Polymyositis
- Inclusion body myositis
- Other systemic autoimmune diseases

Polymyositis & Dermatomyositis

- Polymyositis – disabling muscle weakness
- Dermatomyositis – hyper-pigmentation rashes
- Both occur in 40-60 year olds
- S & S
  - Affects large muscles in shoulders and hips
- Diagnosis
  - Muscle weakness of shoulders and hips in middle age is suggestive, characteristic skin rash
  - EMG, muscle biopsy
- Treatment
  - Steroids and immunosuppressive drugs

Dermatomyositis

- Autoimmune disease affecting muscles and skin
- Symptoms progress over weeks, months
- Rash on the face and neck
- Periorbital edema
- Pain and weakness of proximal limb muscles, neck flexors
- Dysphagia
- Cardiac abnormalities, interstitial lung disease
- Often paraneoplastic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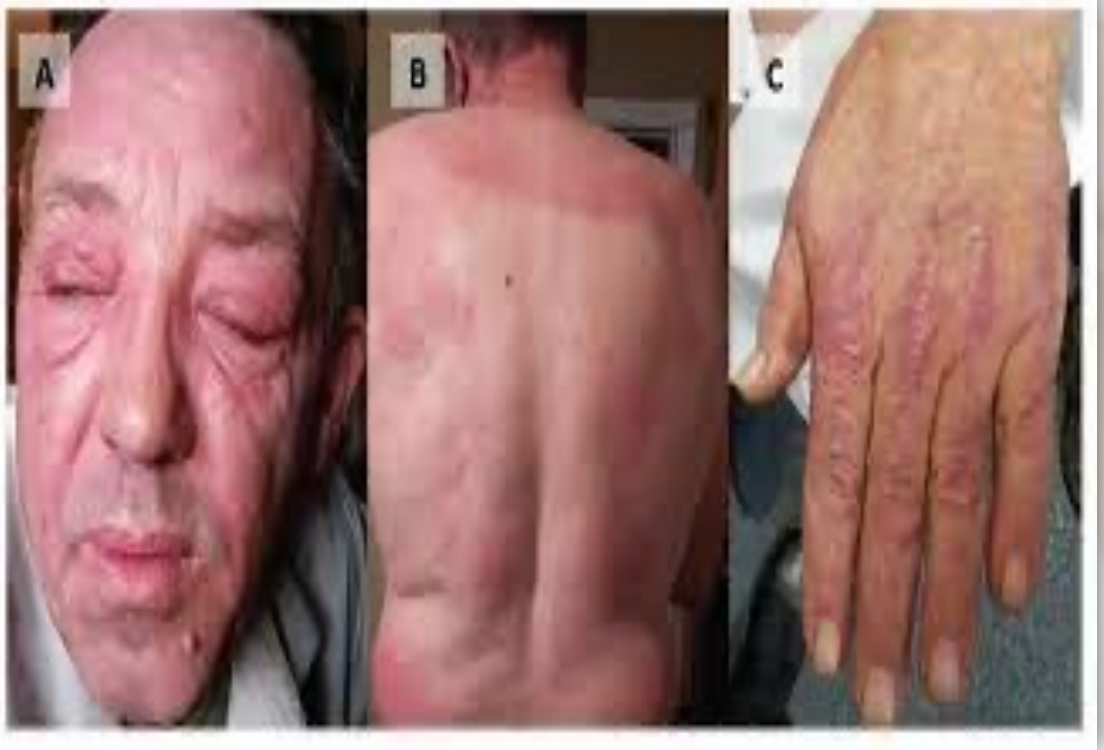
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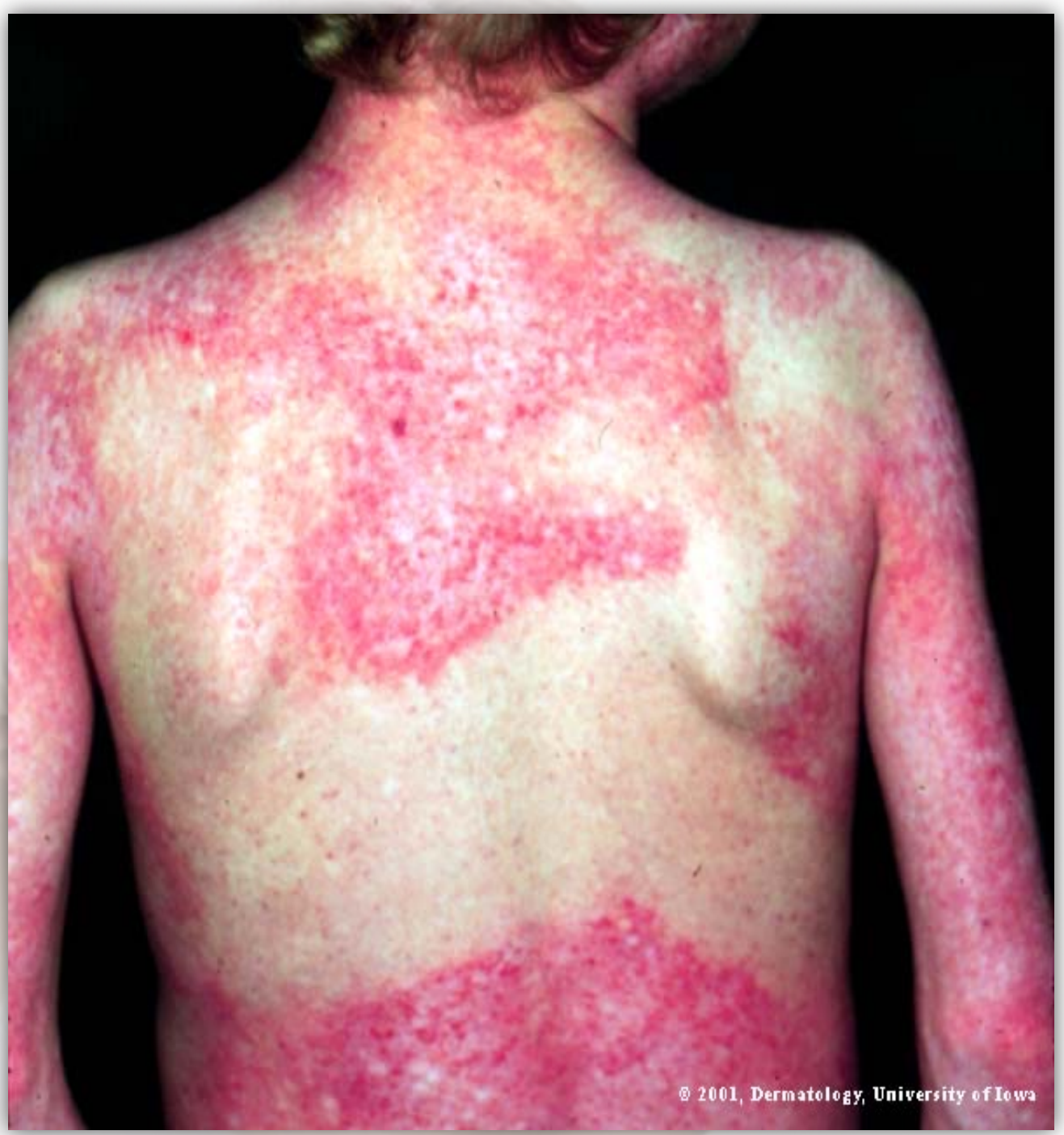
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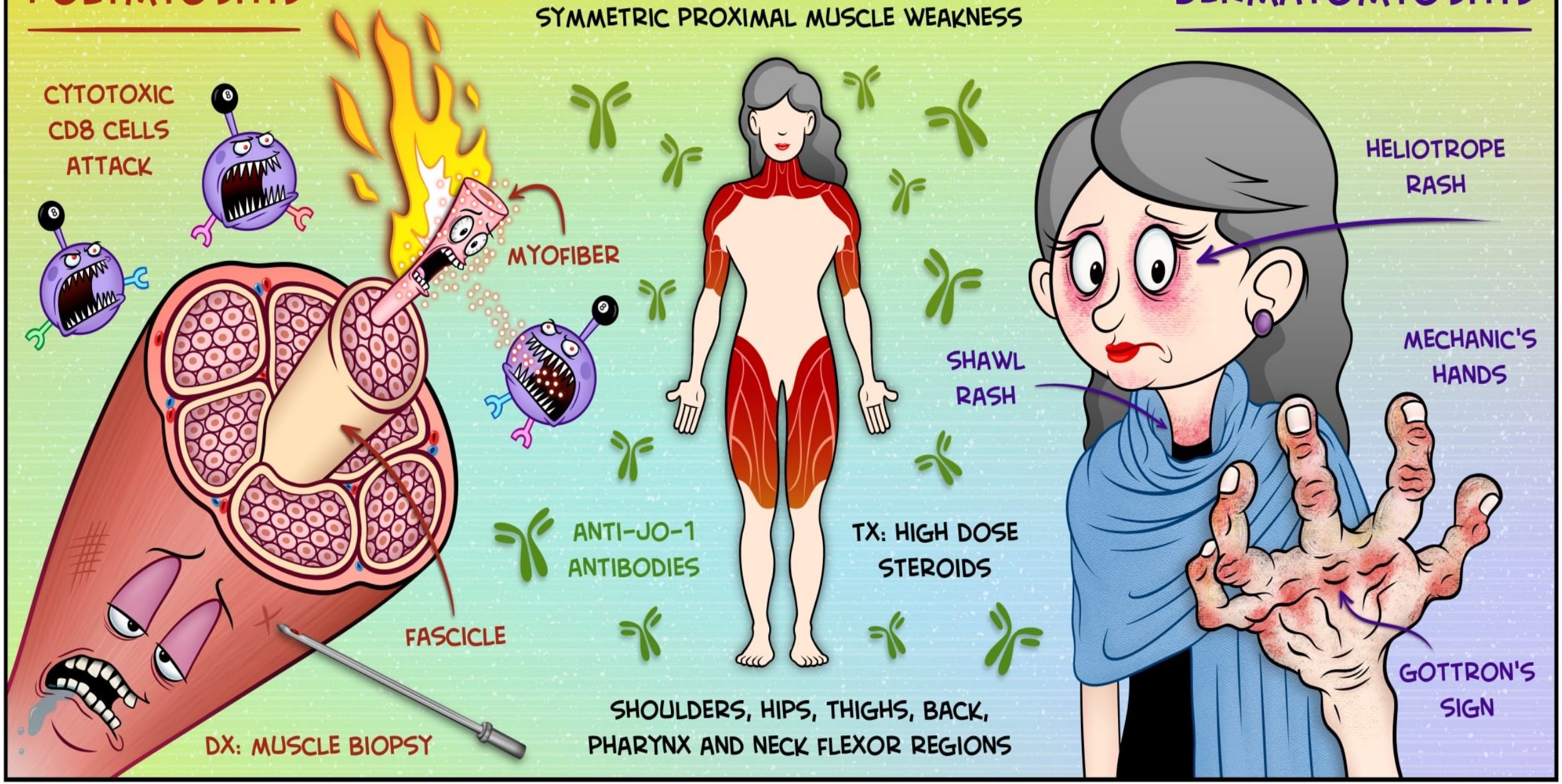
POLYMYOSITIS

INFLAMMATORY MYOPATHIES WITH PROGRESSIVE SYMMETRIC PROXIMAL MUSCLE WEAKNESS

DERMATOMYOSITIS

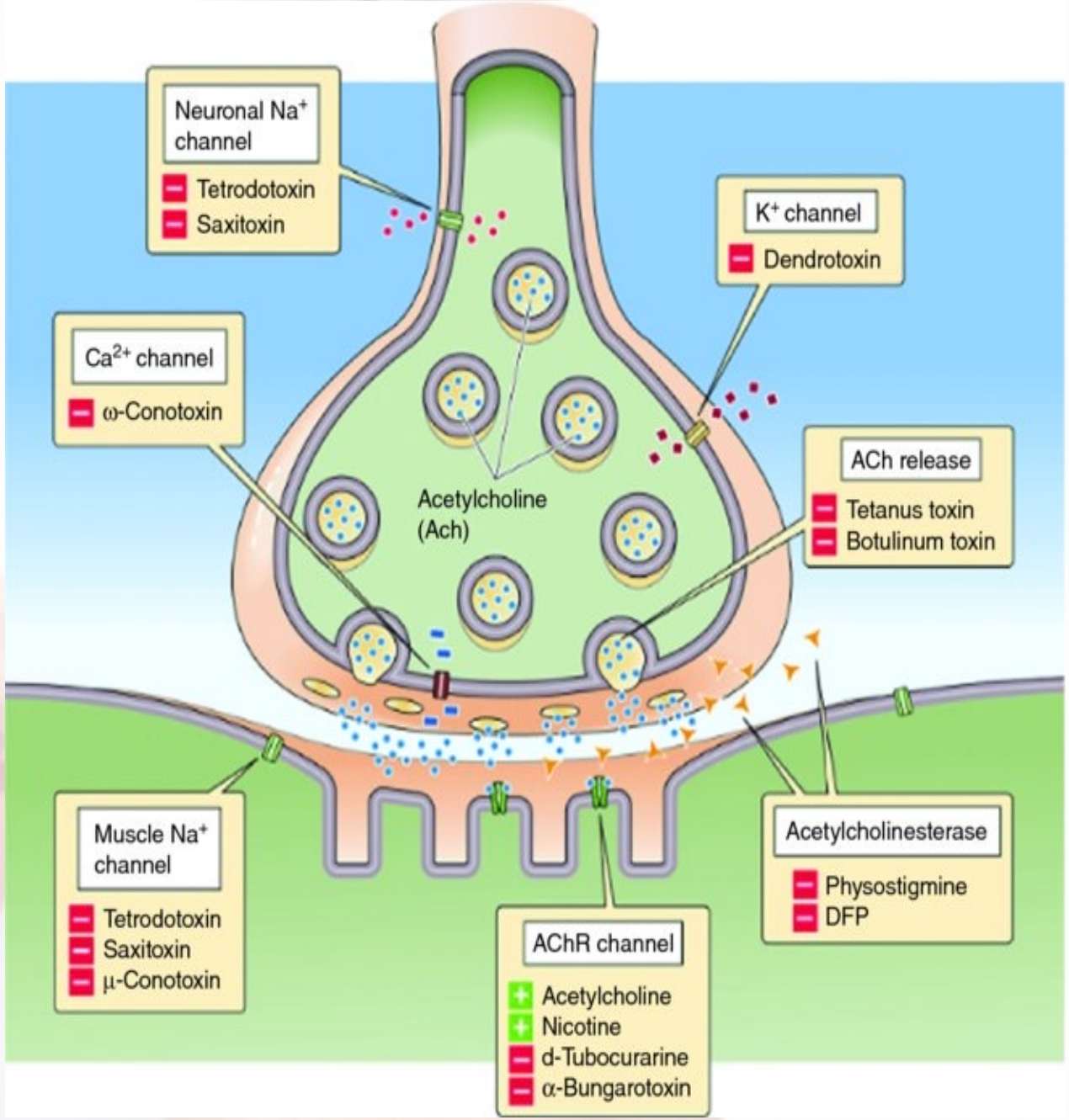


Endocrine and Toxic Myopathies

- Endocrinopathies known to cause myopathy include thyroid and parathyroid diseases, disorders of the adrenal axis such as Cushing syndrome, and acromegaly.
- Patients in the intensive care unit are at risk for developing critical illness myopathy, also known as myosin-loss myopathy, which should be considered if intensive care unit acquired weakness develops.

- Symptoms of endocrine myopathies include weakness and atrophy (shrinking) of the muscles around the shoulders and hips, muscle stiffness, cramps, slowed reflexes, and in severe cases, muscle breakdown.
- Myopathies usually don’t cause muscles to die; instead, they keep them from working properly.
- Myopathies are usually nonprogressive.
  - A myopathy usually doesn’t grow worse over a person’s lifetime.
  - In fact, some children with myopathies gain strength as they grow older.

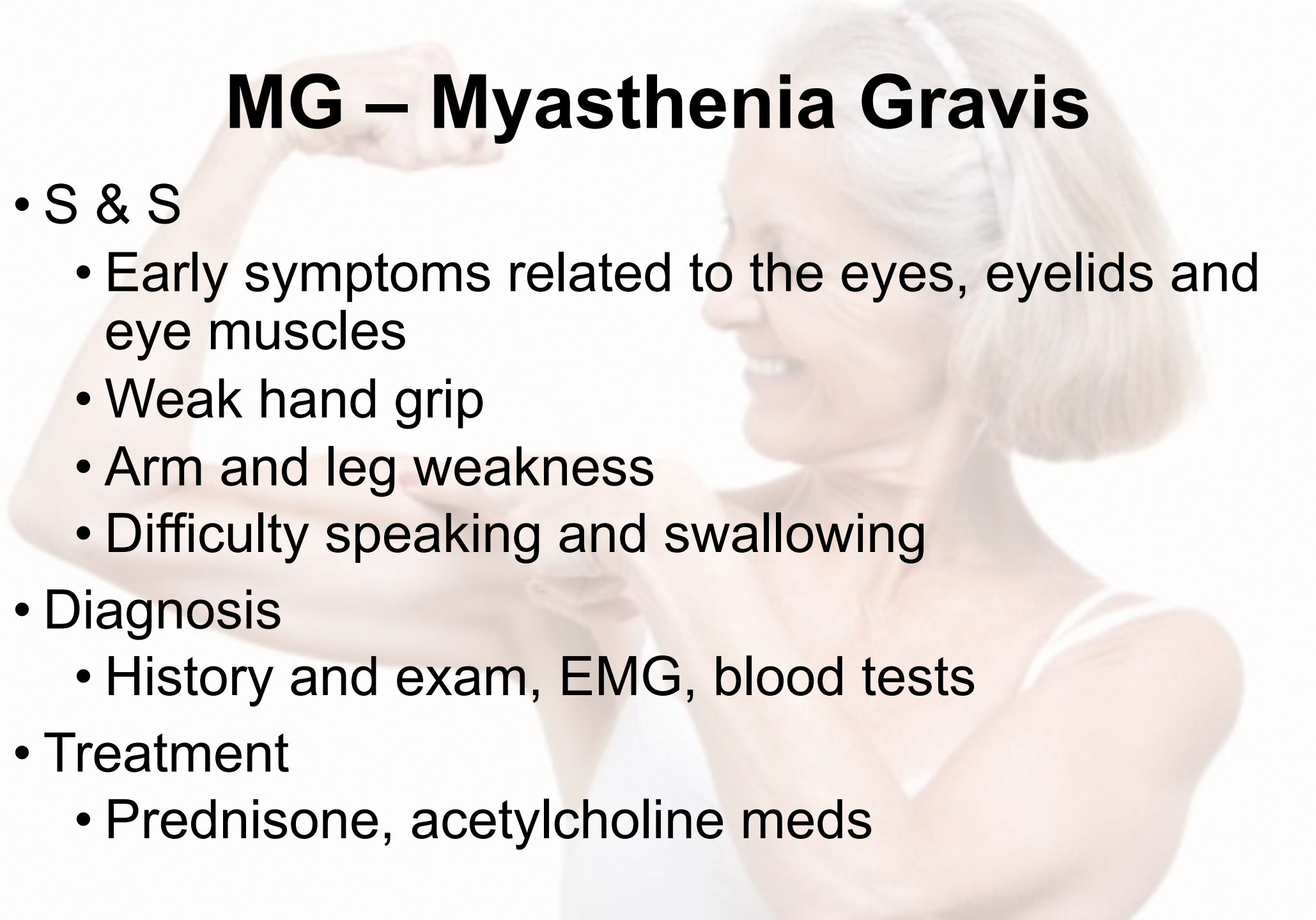
Disorders of the Neuromuscular Junction



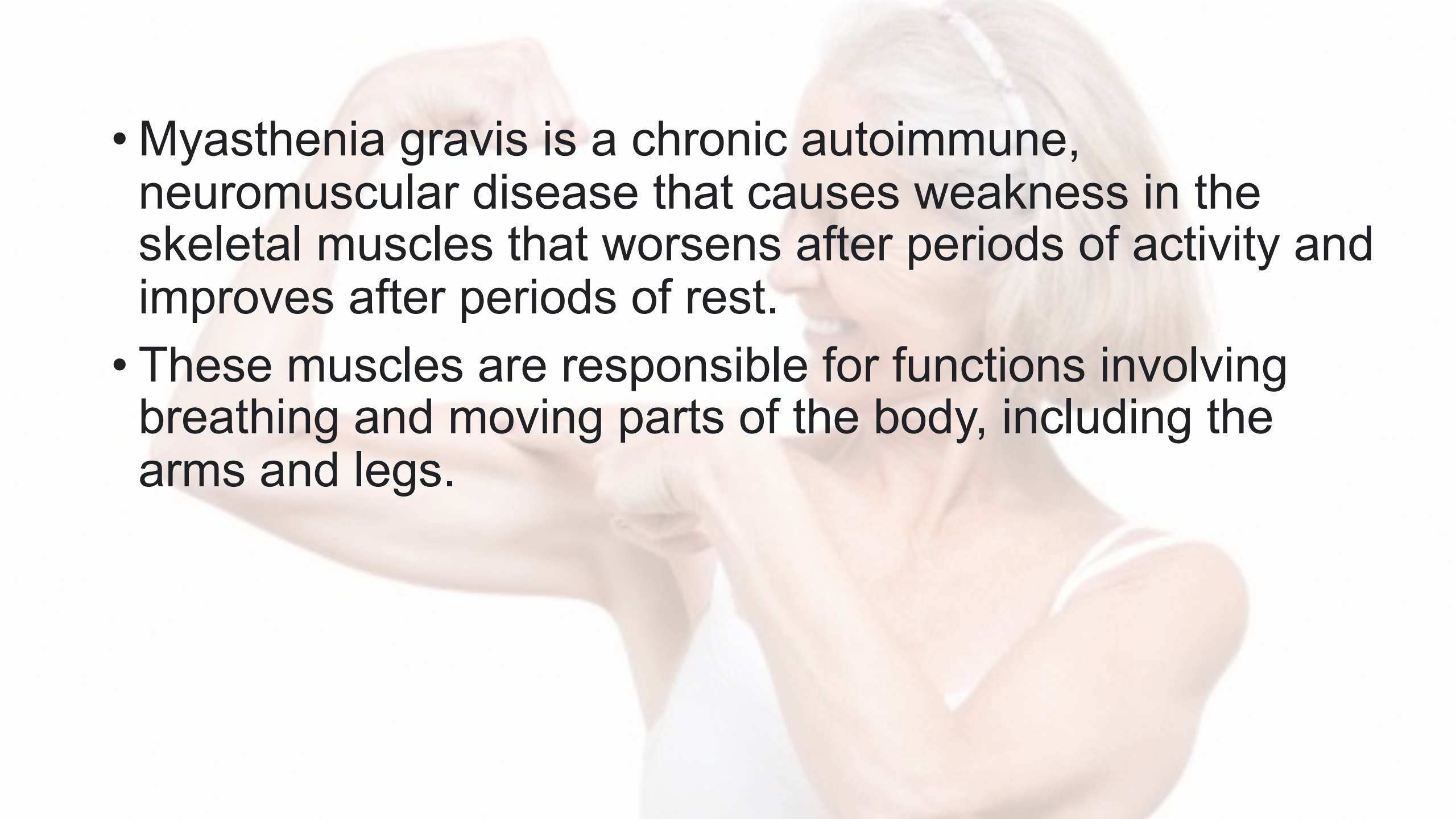
MG – Myasthenia Gravis

- Progressive weakness, an autoimmune disease leading to dysfunction of neuromuscular junction
- Antibodies attack the acetylcholine receptors of the motor end plate of the muscles
- Results in LMN dysfunction with progressive weakness
- More common in women

MG – Myasthenia Gravis



- S & S
  - Early symptoms related to the eyes, eyelids and eye muscles
  - Weak hand grip
  - Arm and leg weakness
  - Difficulty speaking and swallowing
- Diagnosis
  - History and exam, EMG, blood tests
- Treatment
  - Prednisone, acetylcholine meds

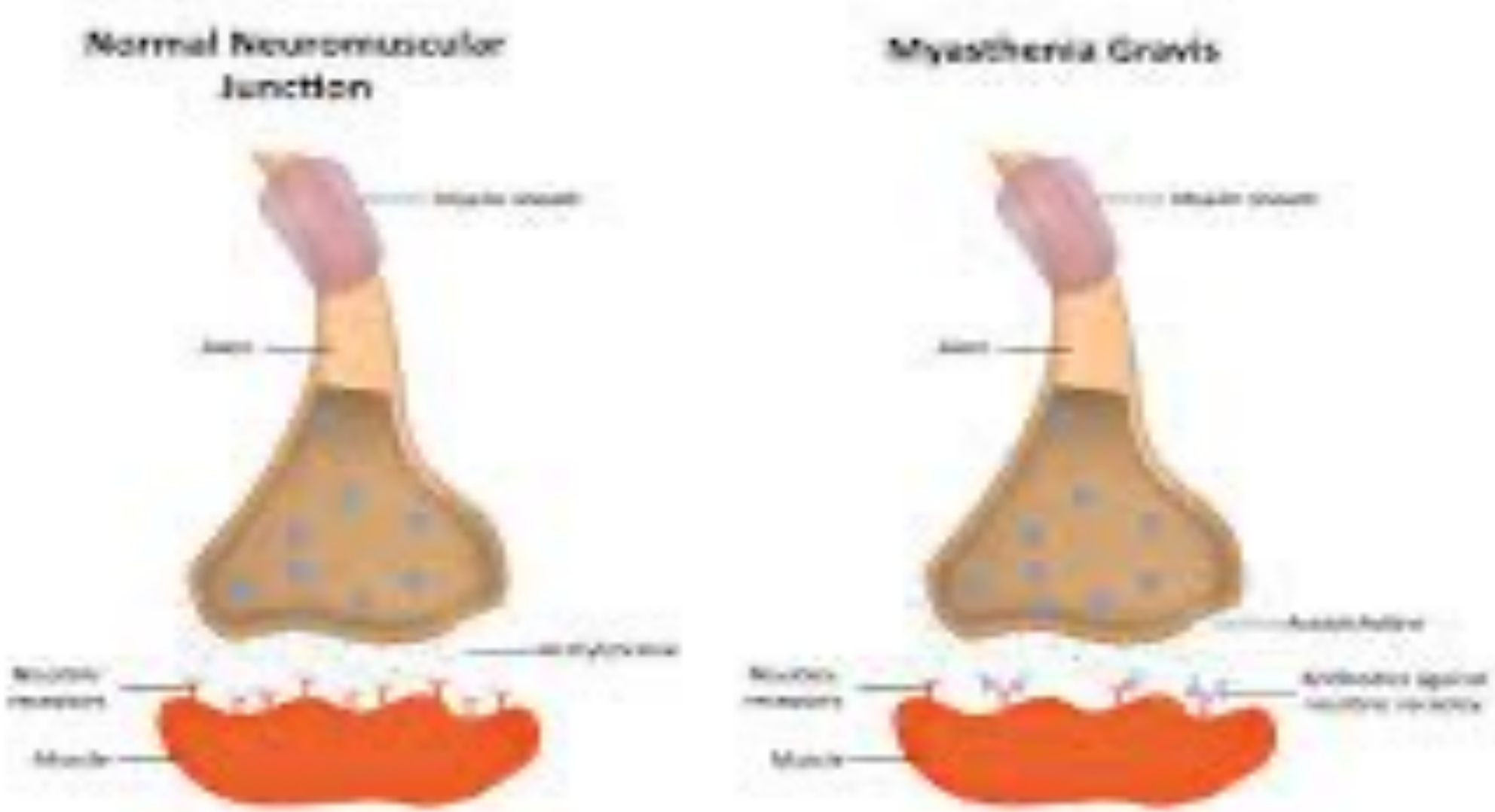


- Myasthenia gravis is a chronic autoimmune, neuromuscular disease that causes weakness in the skeletal muscles that worsens after periods of activity and improves after periods of rest.
- These muscles are responsible for functions involving breathing and moving parts of the body, including the arms and legs.

Muscular Dystrophy Epidemiology

- Incidence 1 : 20 000 in USA
- Women slightly higher incidence 3 : 2
- Majority of the MG are young women in the third decade and middle-aged men in the 5^{th} and 6^{th} decade
- Children account for 11% of all patients

Myasthenia Gravis

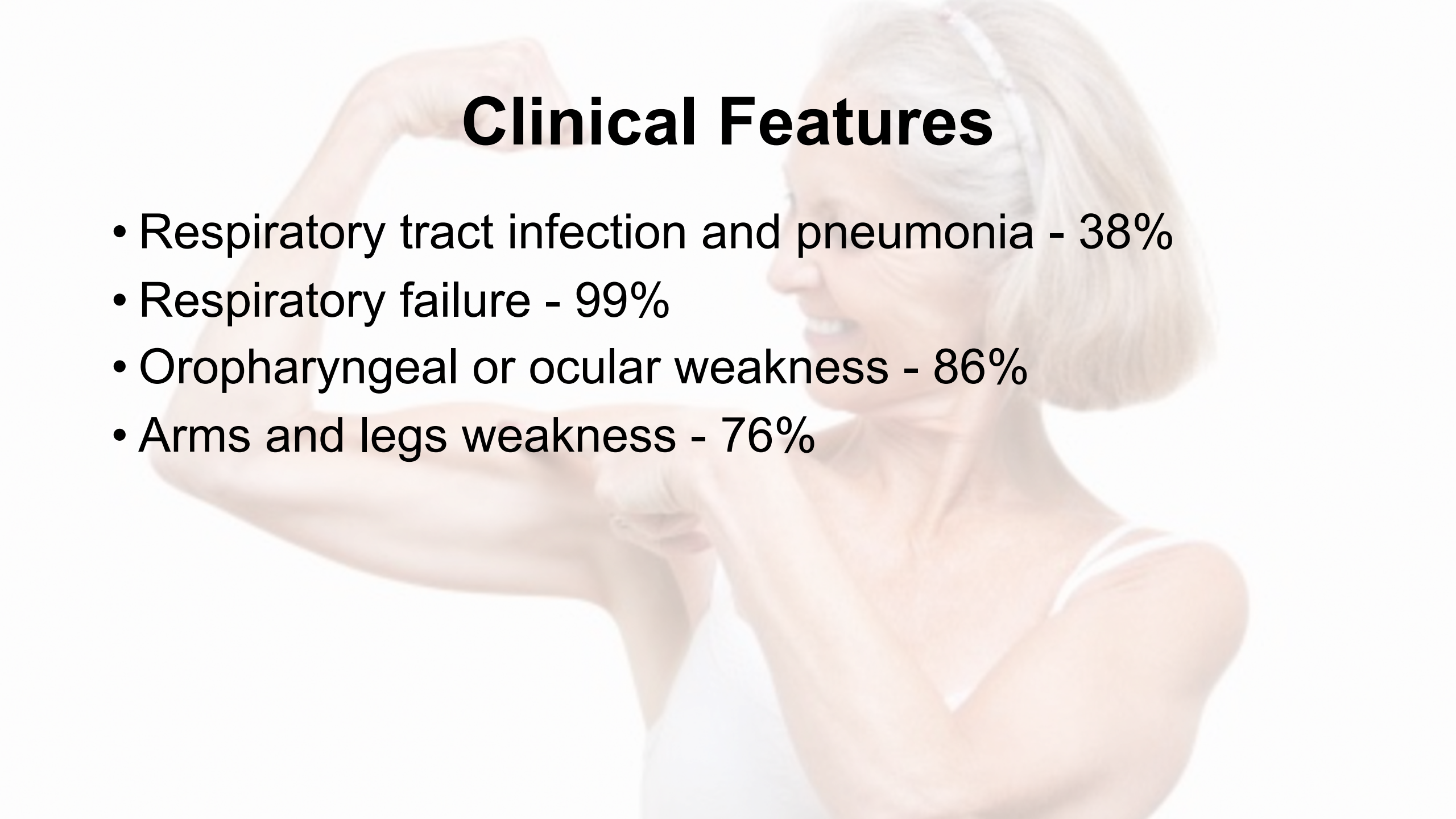


Myasthenic Crisis

- Respiratory insufficiency – paralysis of respiratory muscles
- Assisted ventilation required
- Affect 15-20% myasthenic patients
- Females : males = 2 : 1
- Average age : 55 years

Clinical Features

- Respiratory tract infection and pneumonia - 38%
- Respiratory failure - 99%
- Oropharyngeal or ocular weakness - 86%
- Arms and legs weakness - 76%



Complication of Crisis

- Ateletactic pneumonia - 40%
- Hypotension
- Cardio-respiratory arrest
- Pneumothorax

Treatment of Myasthenic Crisis

- ICU is required for assisted ventilation
  - Usually intubated for 14 days
  - Usually hospitalized 35 days
- Cardiopulmonary monitoring
- Plasmapheresis
- Antithrombotic treatment
- Antibiotics
- Respiratory rehabilitation
- 40% of patients have tracheostomy