

Neuromuscular Disease

Clinical presentations, signs and symptoms

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COI

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Is it neuromuscular ?

Combinations ?

Case history

Clinical presentation

new concepts

examples

Neuromuscular System (A common construct)

- (Extracranial parts of the Cranial nerves)
- Nerve Roots
- Plexus
- Peripheral nerves
- Neuromuscular junction
- Muscle
- Autonomic system

Neuropathies: How often ?

Median (CTS)	103,00
Ulnar	25,00
Radial	2,50
Cut. Femoral	10,00

Disc (> 50 yrs)	
Cervical (aged)	600,00
Lumbar (aged)	1079,00

PNP DM II: 6400

Pro 100 000 Patient years

Source: Callaghan, Ann Arbor 2016, USA

Combinations of neuromuscular and central nervous system. Examples:

- MND/ALS
- Some hereditary neuropathies
- Myotonic dystrophy
- etc.

Case history



Family history (tree)

Diseases, medications,

Exposition, trauma

**Onset, duration, acute, chronic
progressive , fluctuating,
episodic**

Symptoms:

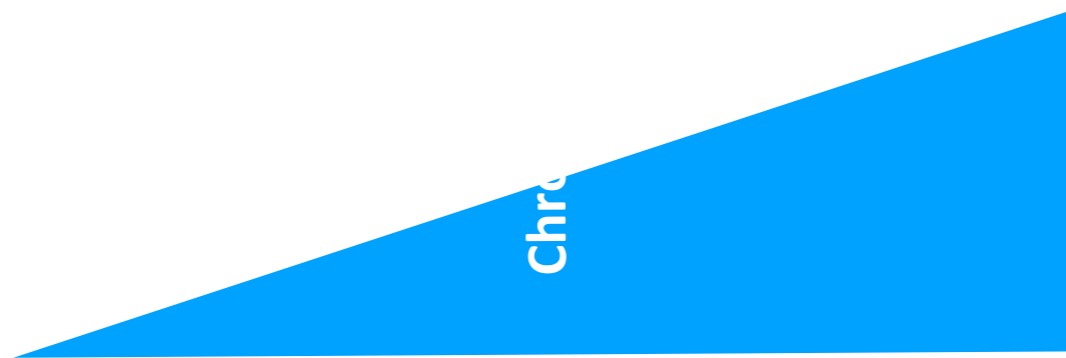
**motor, sensory, autonomic
movement, composite**

Acquired/genetic

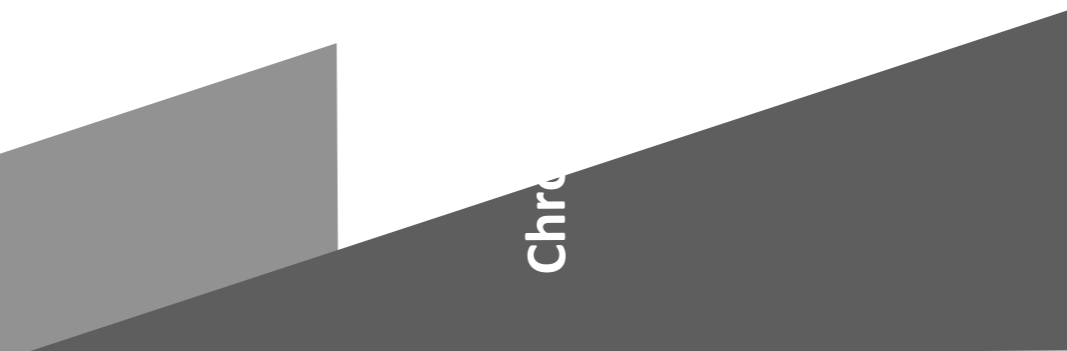
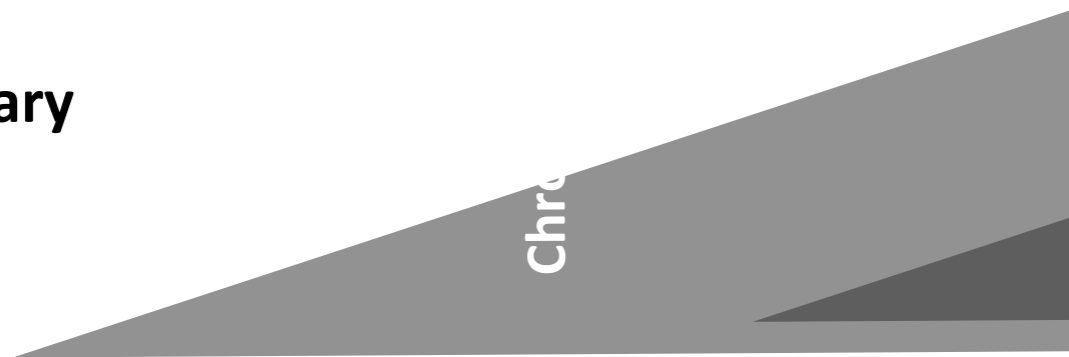
**Before examining the patient-
create hypothesis**

Time

Acquired



Hereditary



Symptoms and signs

Motor

Sensory

Composite function

Autonomic

Neuropathic Pain

Distribution

Symptoms and signs

Skin

Skeletal system

Motor

Distribution of weakness, atrophy

Strength (BMRC)

Time: Fluctuating, episodic

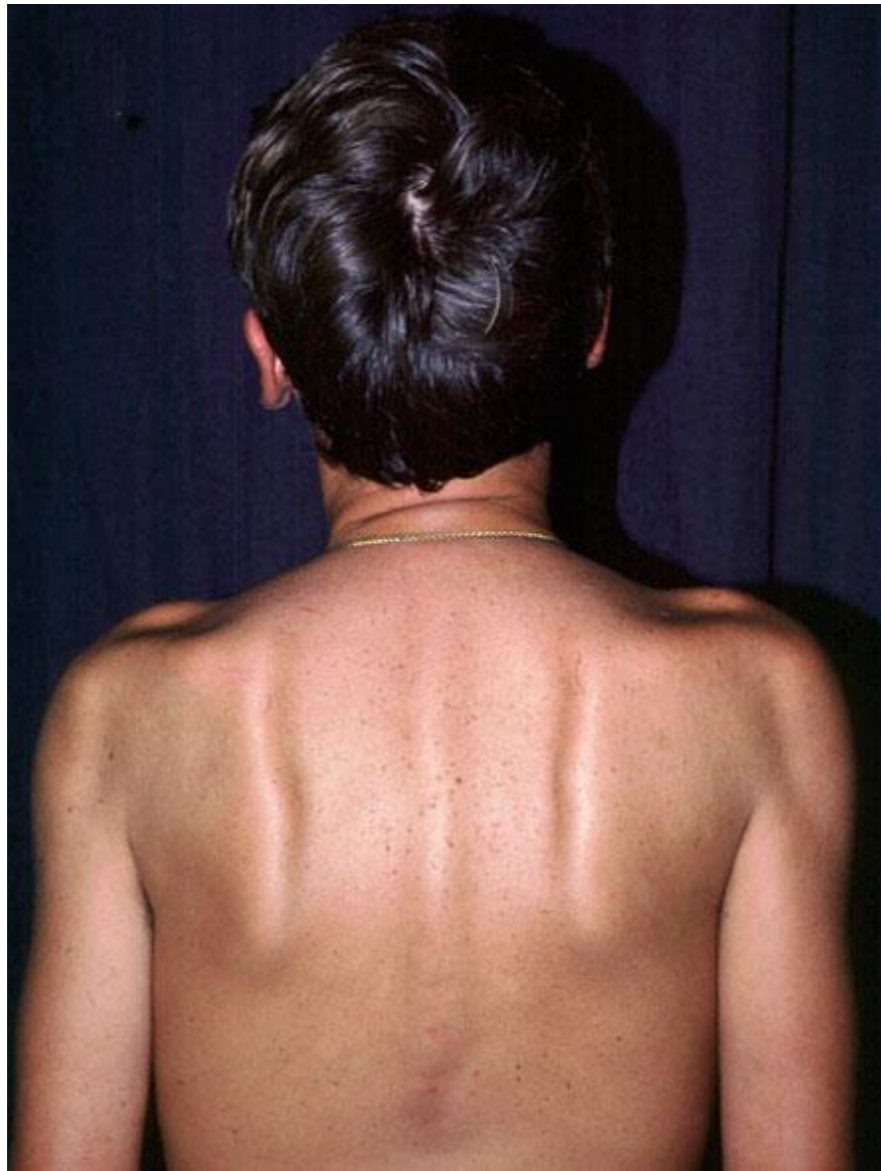
Muscle volume

Atrophy

Hypertrophy

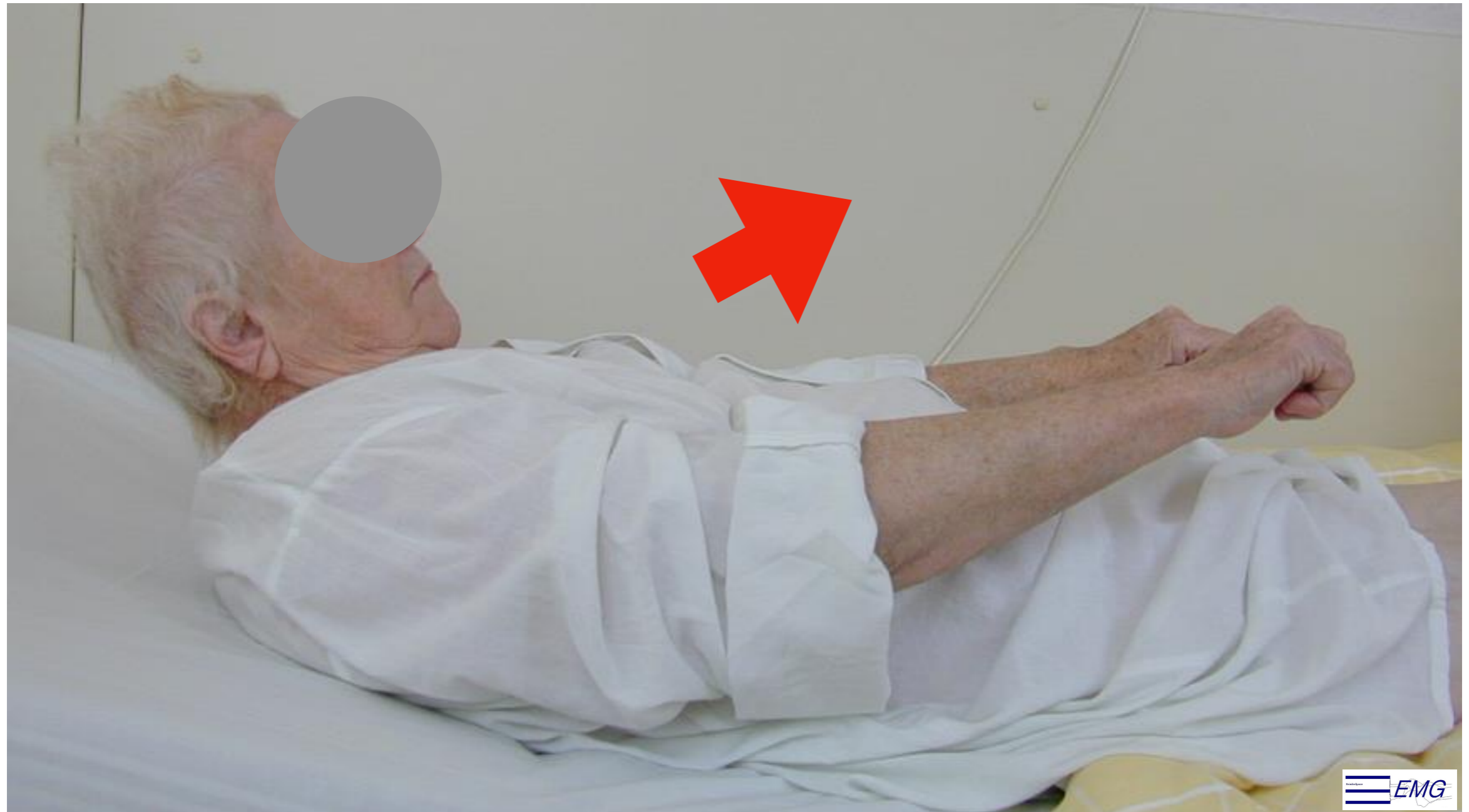
Phenotype distribution: myopathy, polio, club foot

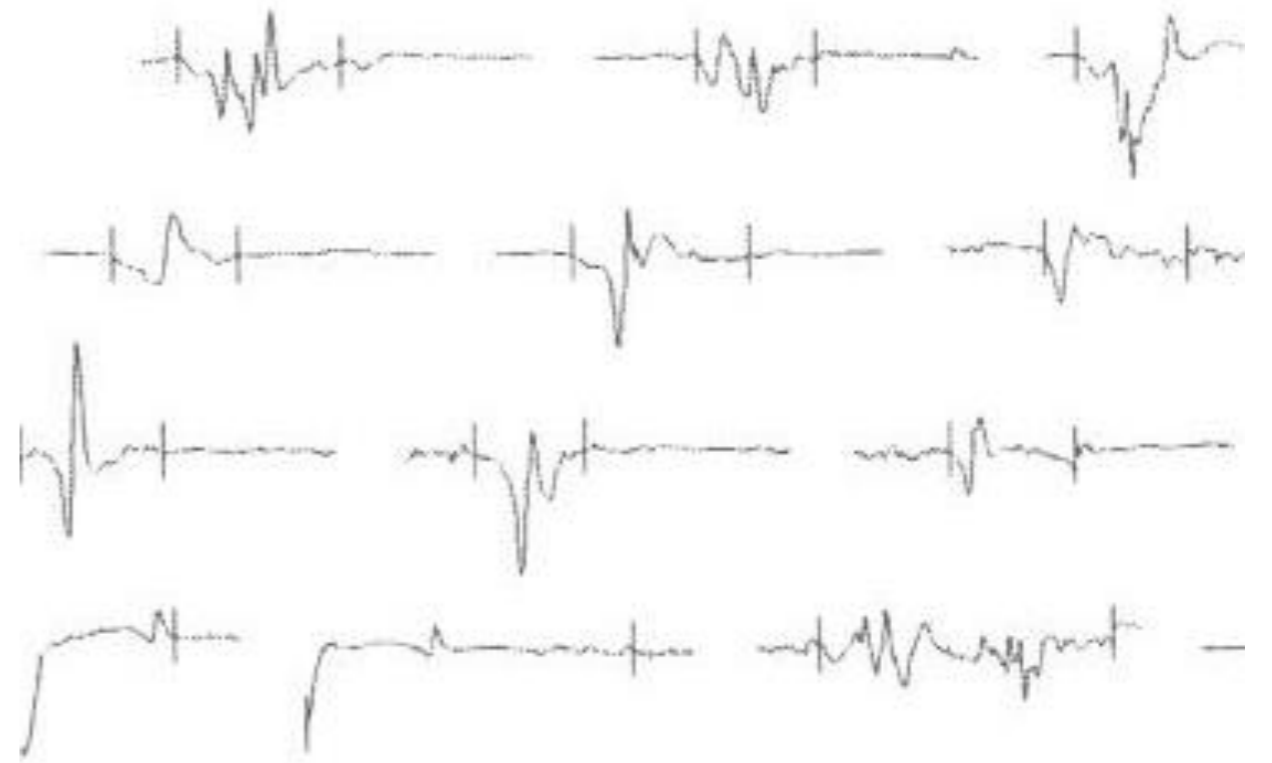
Winging





Proximal weakness





Carcinomatose Neuromyopathie

EMG

Percussion myotonia



Muscle hypertrophy

Examples

Pseudohypertrophy

Endocrinopathy

Focal muscle hypertrophy

Overuse

infections

Amyloid

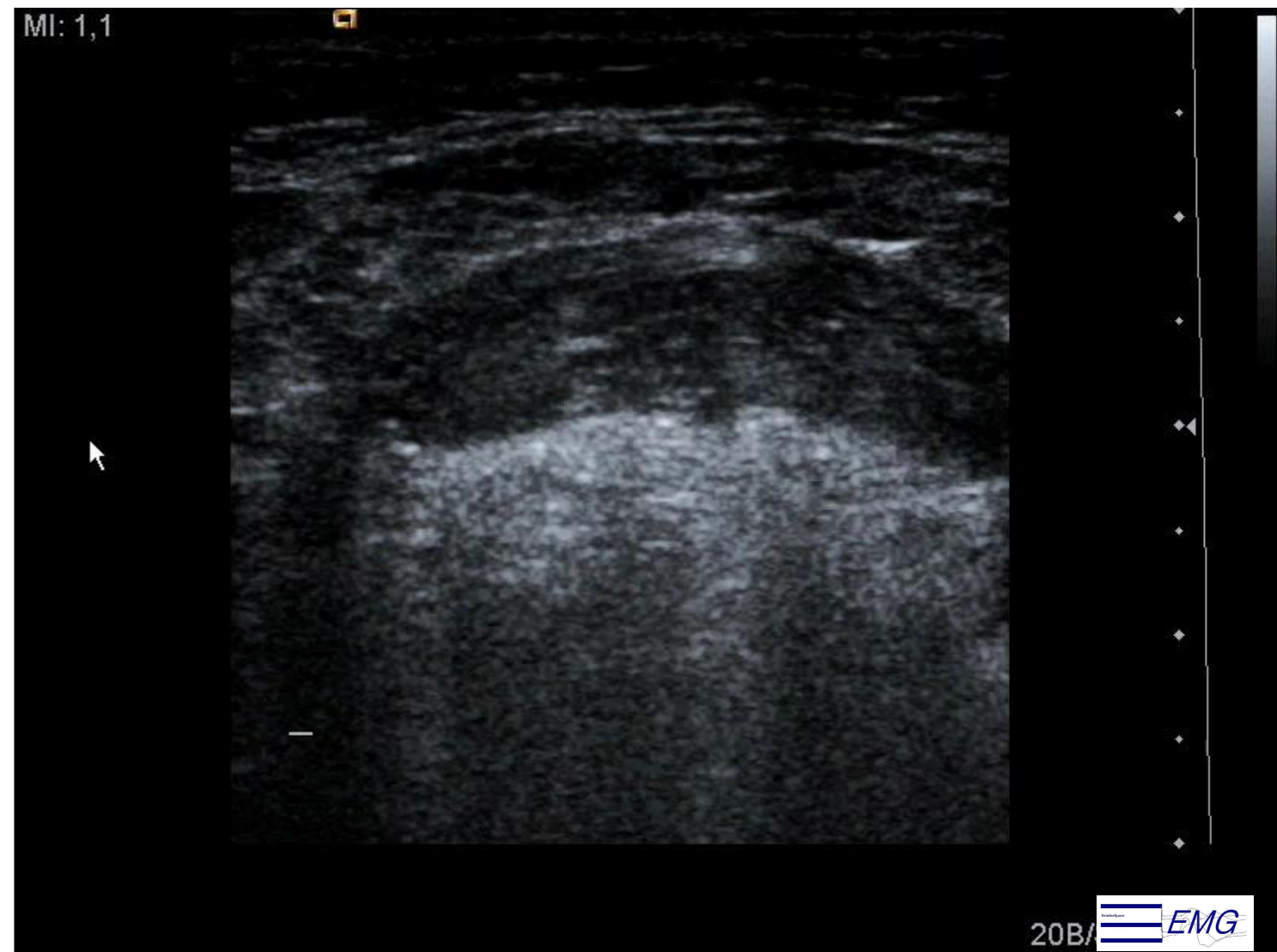


Steroid myopathy



Movement

- Fasciculation
- Myokymia
- Rippling
- „Stiffness“, myotonia
- Percussion myotonia
- Cramps



Tongue Ultrasound

Sensory

Quality

Sensory assessment

Instruments

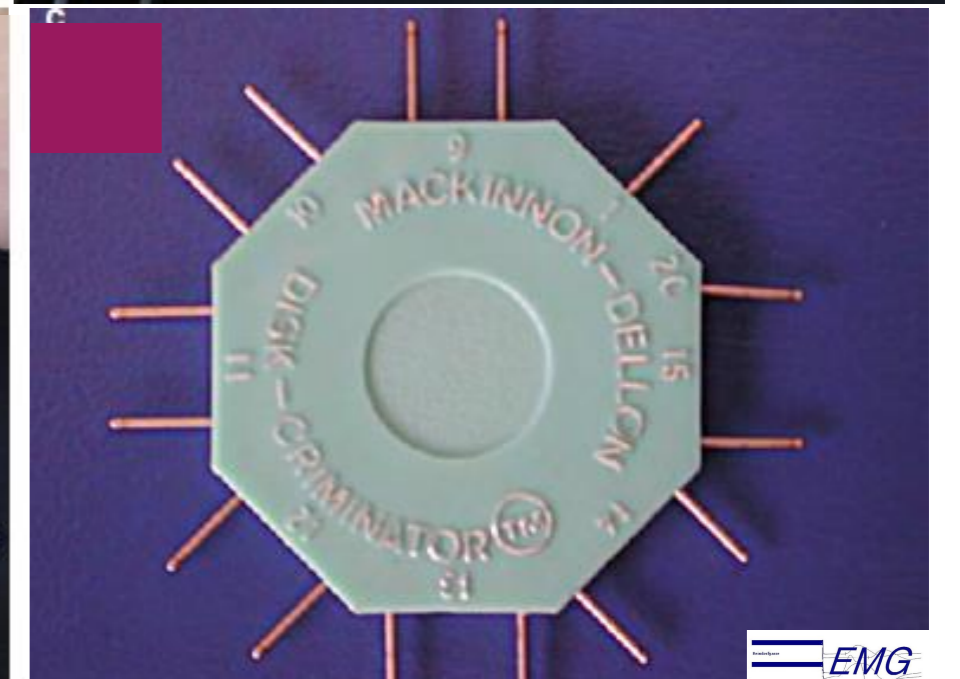
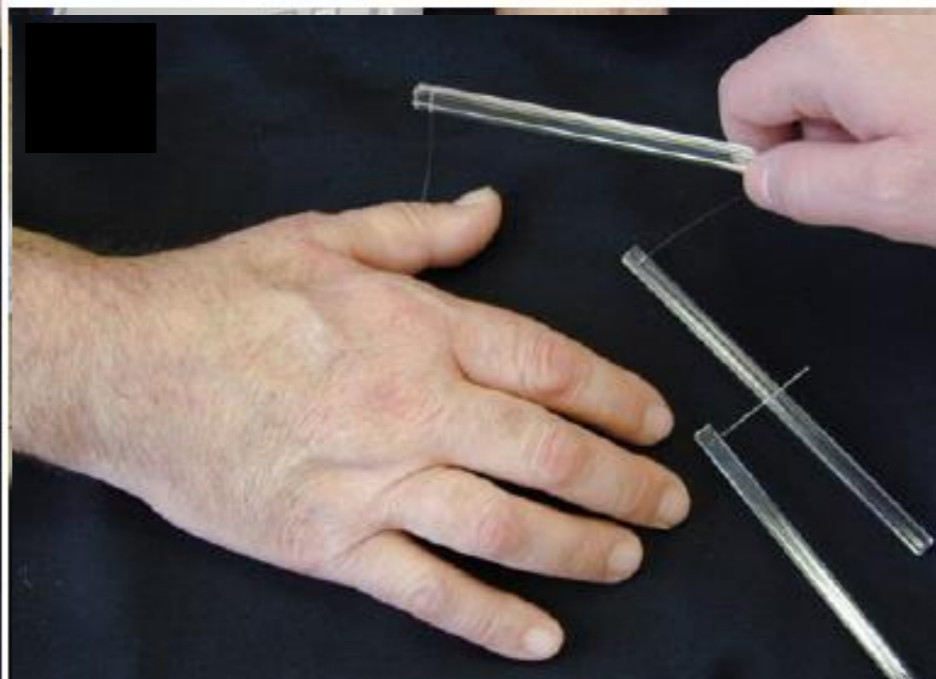
Type of fiber lesion

Tinel Hofmann sign

Sclerotoma,
myotoma,
„neurotoma“



Instruments





A



B

A: superficial peroneal nerve

B. Infrapatellar nerve

Tinel Hofmann: red circle



Foot

Test Composite function !

Simple (but complex) functions:

Opening and closing buttons

Recognition of objects

Balance: Romberg

Stance on one leg

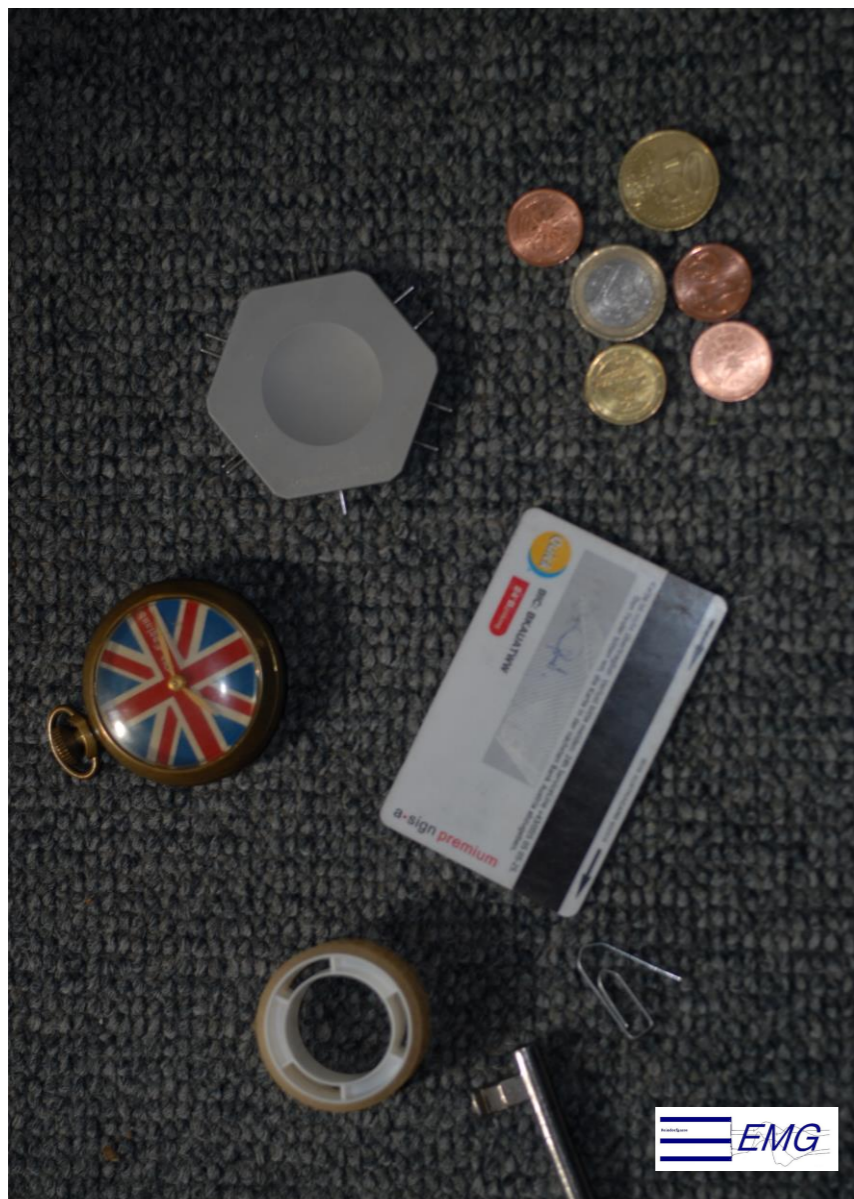
Can you put on your trousers standing
freely one leg ?

Muscle:

„Hands up“

Standing up from squat

Push ups



**Ask for
Autonomic
dysfunction**

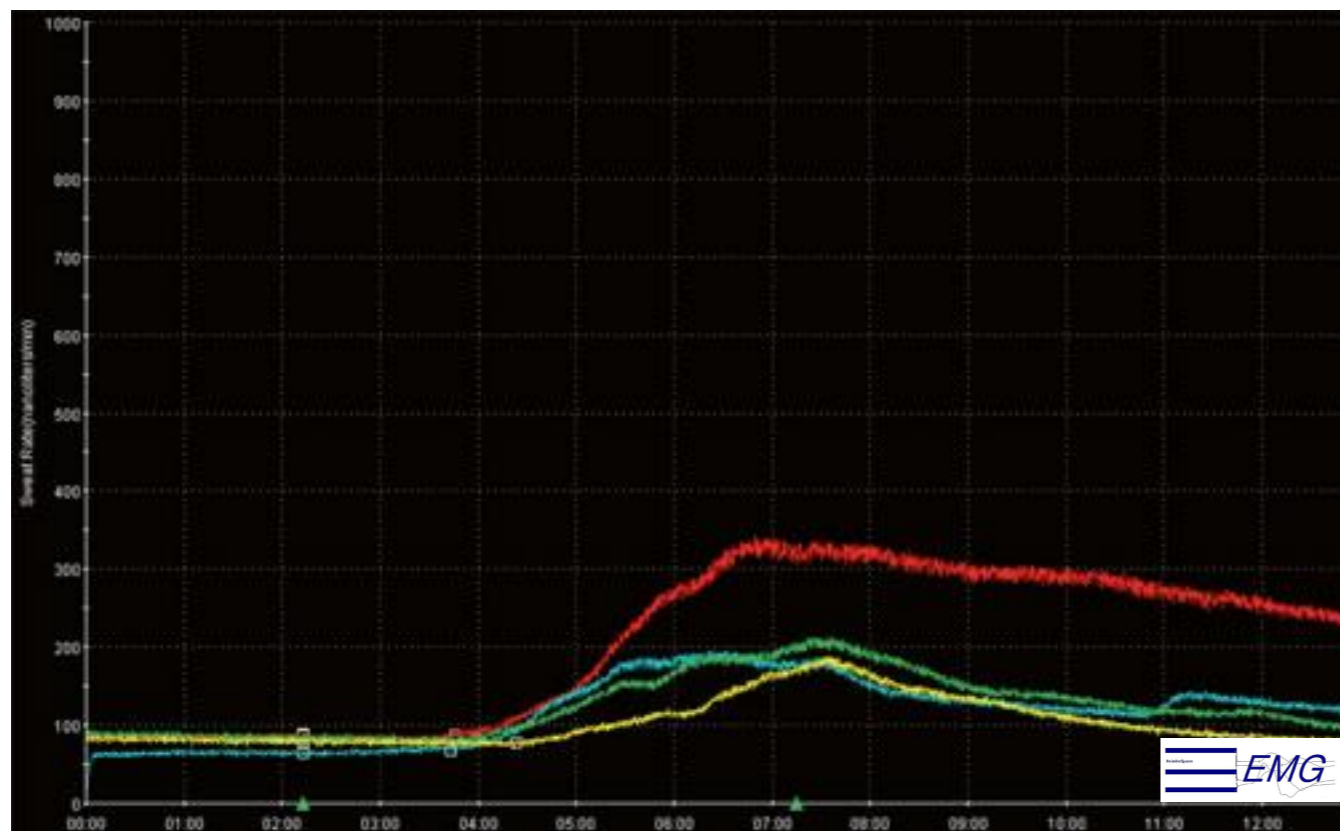
Dry mouth, dry eyes ?

Voiding

Sexual dysfunction

Orthostasis

Gastrointestinal



Neuropathic Pain

Pain characteristics

(stabbing, shooting, electric)

Distribution- cave Allodynia

Sensory, motor involvement

Elicited by : eg Tinel Hofmann

Neuropathic Pain conditions

- Acute episodic
- Painful neuropathy
- Small fiber
- Trauma causalgia
- Localized
- Surgery
- Erythromelalgia
- Phantom- pain
- PRURITUS

Intercostobrachial nerve



Painful neuropathies

- Alcohol
- Metals
- Drugs (eg. CIPN)
- Immune-mediated, Vasculitis
- Hereditary (eg Fabry)

Skin

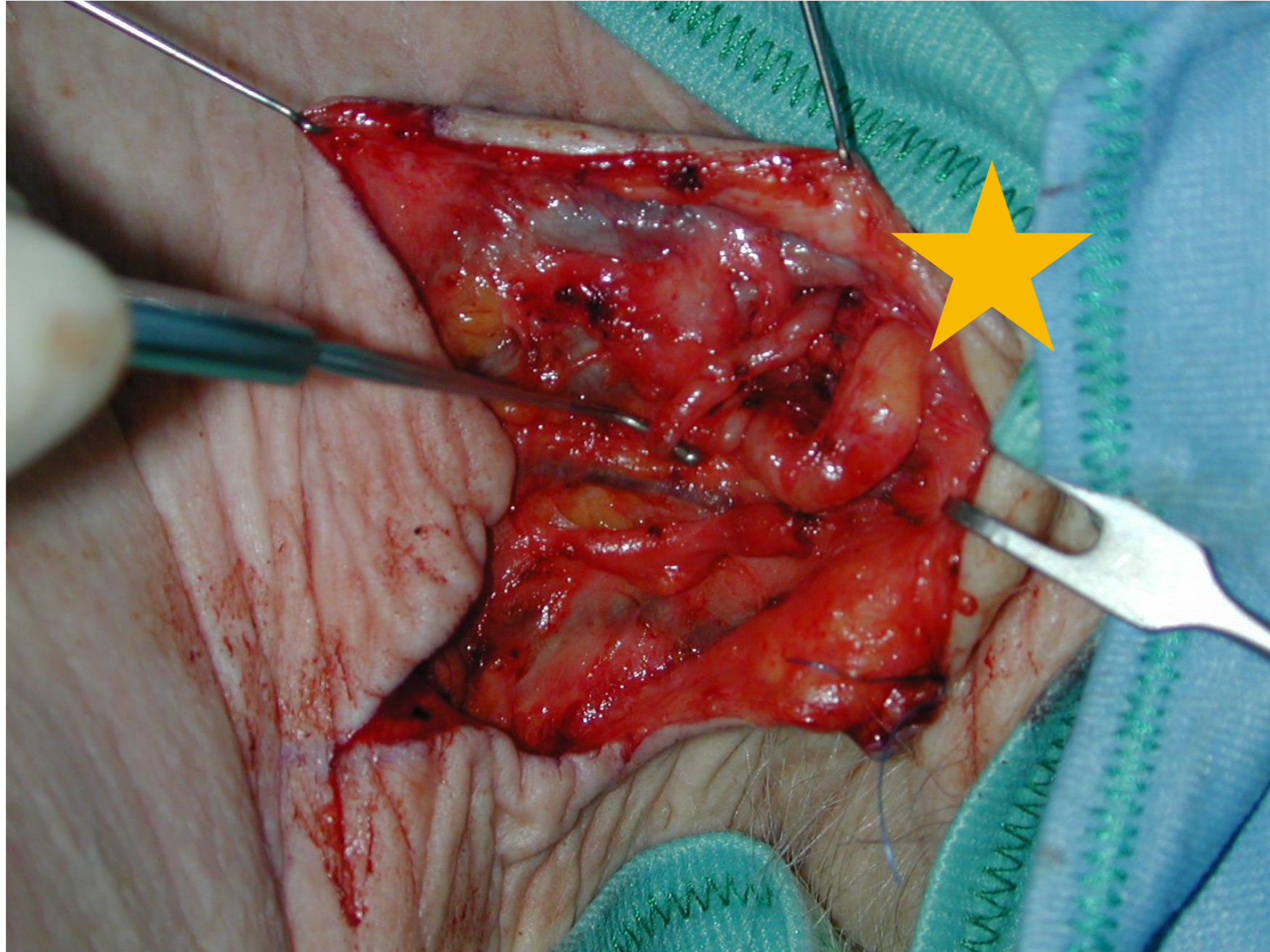


Haemangioma



Waterhouse F. syndrome

Thickening of nerves



Greater auricular nerve (intraoperative)

Radiofibrosis syndrome





Hyperkeratosis in denervation



Skin



Lymphoma

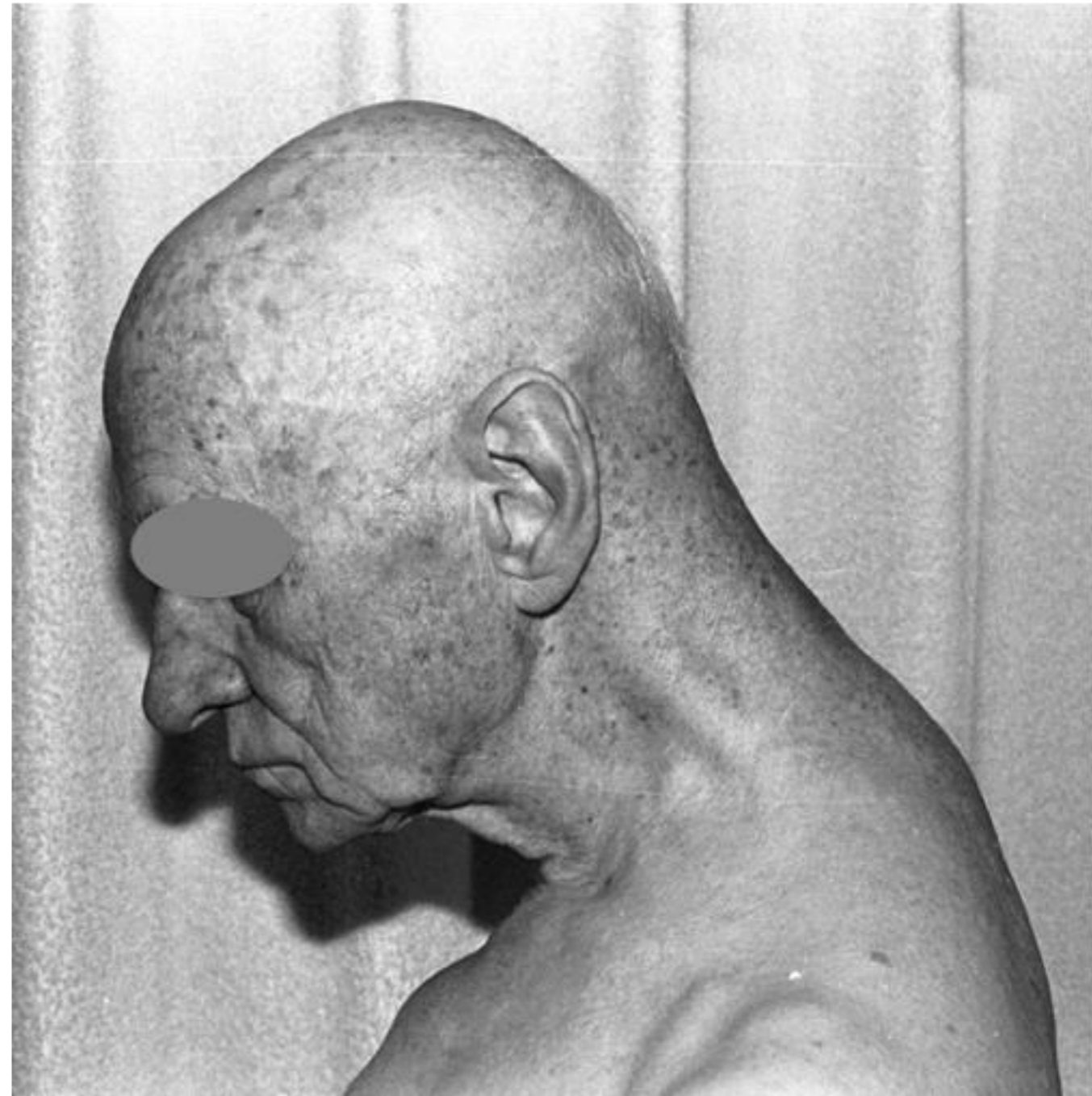
Hand Foot Syndrome



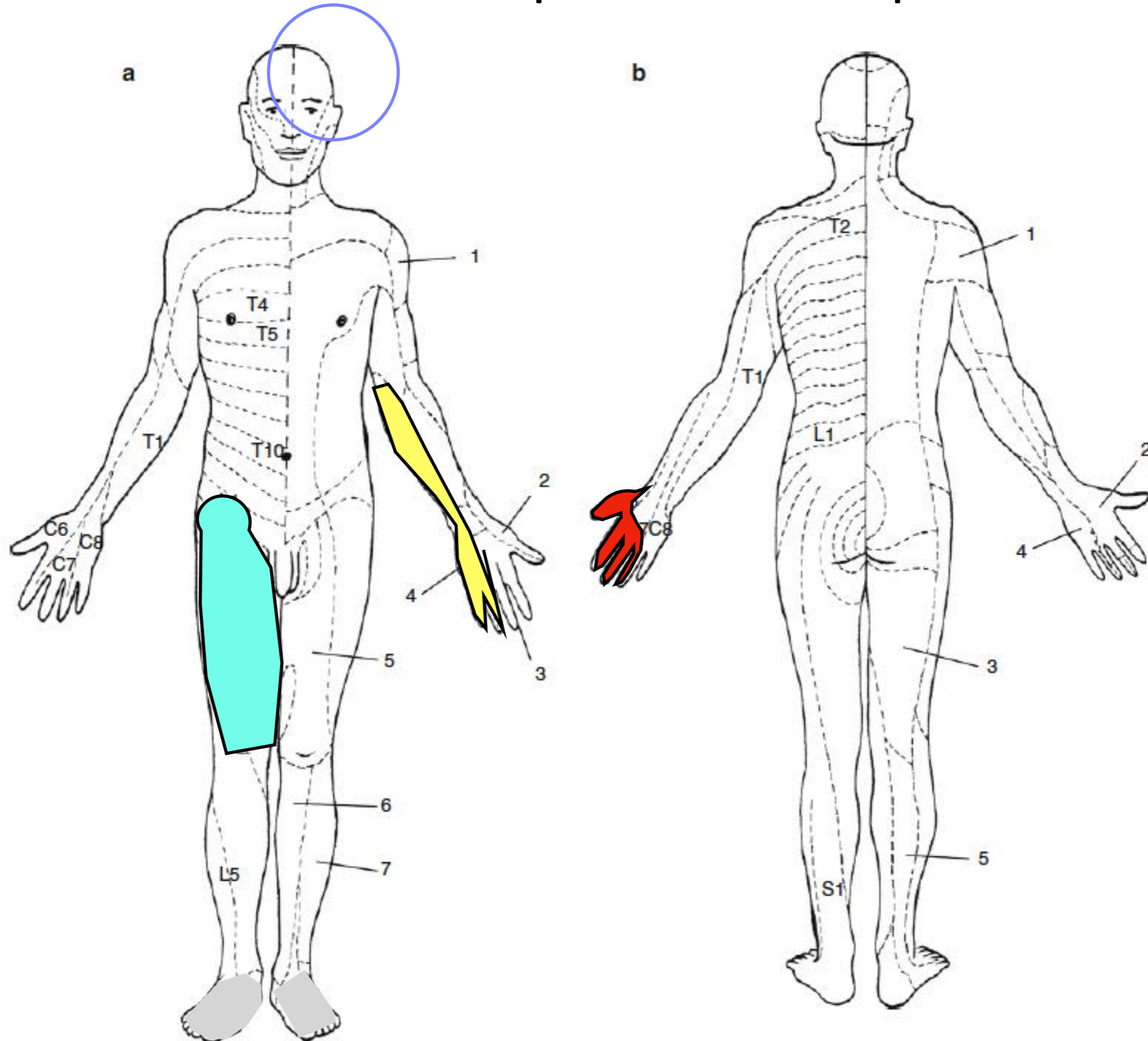
Skeletal system



Focal dysfunction/Weakness



Distribution: Neuropathies and Radiculopathies



Further Ancillary Tests

- NCV/EMG
- Imaging: ultrasound, MRI, CT
- Autonomic test
- Biopsy
- Laboratory
- Genetics

Nerve Ultrasound



Neuromuscular lesions

- CN
- Plexus lesions
- Mononeuropathies
- Polyneuropathies
- Neuromuscular transmission
- Muscle