Neuromuscular Disease
Clinical presentations, signs and symptoms

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Neuromuscular System (A common construct)

- (Extracranial parts of the Cranial nerves)
- Nerve Roots
- Plexus
- Peripheral nerves
- Neuromuscular junction
- Muscle
- Autonomic system
### Neuropathies: How often?

<table>
<thead>
<tr>
<th>Neurological Pathology</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Median (CTS)</td>
<td>103,00</td>
</tr>
<tr>
<td>Ulnar</td>
<td>25,00</td>
</tr>
<tr>
<td>Radial</td>
<td>2,50</td>
</tr>
<tr>
<td>Cut. Femoral</td>
<td>10,00</td>
</tr>
<tr>
<td>Disc (&gt; 50 yrs)</td>
<td></td>
</tr>
<tr>
<td>Cervical (aged)</td>
<td>600,00</td>
</tr>
<tr>
<td>Lumbar (aged)</td>
<td>1079,00</td>
</tr>
</tbody>
</table>

**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 Neuromyopathy

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“
A: superficial peroneal nerve

B. Infrapatellar nerve

Tinel Hofmann: red circle
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 Ancilliary Tests

• NCV/EMG

• Imaging: ultrasound, MRI, CT

• Autonomic test

• Biopsy

• Laboratory

• Genetics
Nerve Ultrasound
Neuromuscular lesions

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