Clinical Aspects of Peripheral Nerve and Muscle Disease

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1. Anterior Horn Cell
2. Dorsal root ganglion cell
3. Motor Peripheral Nerve
4. Sensory Peripheral Nerve
5. Neuromuscular Junction
6. Sensory ending
7. Muscle
Peripheral Neuropathy

- Mononeuropathy
- Mononeuropathy Multiplex
- Polyneuropathy
- Distal distribution of Sensory and motor signs and symptoms
Peripheral Neuropathy

1. Sensory:
   a. Paraesthesia (tingling, pins & needles, prickling, burning...)
   b. Numbness (Hypoaesthesia)
   c. Abnormal degrees and types of sensation

2. Motor:
   a. Weakness
   b. Wasting
   c. Muscle twitching (fasciculations)
Peripheral Neuropathy Investigations

• Blood tests: Part of the general investigation
• CSF:
  – Protein raised in inflammatory conditions
• Radiology (CXR, MR Imaging
• Nerve Conduction Studies + EMG
  – Axonal degeneration Vs Demyelinating
MRI of cervical Nerve root tumours
MRI of cervical Nerve root tumour
Peripheral Neuropathy

Investigations

• Nerve Biopsy
  – Axonal degeneration
  – Segmental Demyelination
  – Inflammatory changes
  – Others (Amyloid, paraprotein,..)
Axonal Degeneration and Regeneration
Segmental Degeneration  

Autoimmune Segmental Degeneration
Clinical Approach to Peripheral Neuropathy

• Which systems are involved?
• What is the distribution of weakness?
• What is the nature of the sensory involvement?
• What is the temporal evolution? **Acute, Subacute, Chronic, relapsing**,..
• Is there evidence of heredity?
Where is the pathology?
Dermatomes
Clinical Evaluation of Compression Neuropathy
Toxic Neuropathies
Nerve Biopsy

• When to biopsy a nerve?
  – Neuropathy is severe, actively worsening
  – Essential for diagnosis

• Complications
  – 10-15%
  – Localised Sensory loss
  – Wound infection, dehiscence, neuroma formation
  – Unpleasant dysaesthesiae, neuropathic pain
Peripheral Neuropathies in Which a Nerve Biopsy May Be Useful

• **Acquired**
  - Vasculitis*
  - Sarcoidosis*
  - Amyloidosis*
  - CIDP
  - IgM Paraproteinaemic N
  - Leprosy
  - Tumour infiltration*

• **Hereditary**
  - CMT types 1A, 1B & 3
  - HNPP (tomaculous neuropathy)
  - Amyloidosis
  - Giant Axonal Neuropathy
  - Metachromatic Leukodystrophy
  - Polyglucosan body N*
  - Refsum’s disease

*Nerve Biopsy often essential for Dx
Muscle disease
Diagnosis of Muscle Disease

Clinical

Pathology

Genetics
Clinical Classification of Myopathies: Primary diseases of Muscle

- **Hereditary:**
  - Muscular dystrophies
  - Myotonias
  - Channelopathies
  - Congenital myopathies
  - Metabolic myopathies
  - Mitochondrial myopathies

- **Acquired:**
  - Inflammatory myopathies
  - Endocrine myopathies
  - Myopathies associated with other systemic illness
  - Drug-induced myopathies
  - Toxic myopathies
Myopathy

- **Features Supporting Diagnosis:**
  - Distribution: proximal
  - Muscle Bulk: preserved or enlarged
  - Reflexes: Parallel muscle strength
Distribution of weakness can aid diagnosis in muscular dystrophies:
a] Duchenne and Becker;
b] Emery Dreifuss;
c] limb-girdle;
d] fascioscapulohumeral;
e] distal;
f] oculpharyngeal

Most myopathies have a proximal distribution of weakness and pain.
Myopathy

• **Features against diagnosis:**
  – Distal weakness
  – Fasciculations
  – Tremor
  – Sensory signs (or symptoms)
  – Pathological fatigue
  – Early absence of reflexes
Muscle biopsy

• Which Muscle?
  – Moderately involved, but avoid muscle with severe weakness
  – Best Specific Muscles: Deltoid, Biceps, Quadriceps
  – Avoid: Muscle sampled by EMG or sites of recent trauma
EM of muscle fiber

Myofibils

Nucleus

Sarcolemma

Basal lamina or Basement membrane

Satellite cell

Macrophages

Regenerating myoblast

Perivascular Lymphocytes

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