Fabry disease

Theoretical usefulness of electrophysiologic evaluation

Olivier Bouquiaux – François Wang
<table>
<thead>
<tr>
<th>Fiber types</th>
<th>LARGE FIBERS</th>
<th>SMALL FIBERS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aβ &amp; motor fibers</td>
<td>5-15 µm</td>
<td>1-5 µm</td>
</tr>
<tr>
<td>Diameter</td>
<td>1-5 µm</td>
<td>0,3-1,5 µm</td>
</tr>
<tr>
<td>Myelin sheath</td>
<td>+++</td>
<td>+</td>
</tr>
<tr>
<td>Conduction velocity</td>
<td>40-100 m/s</td>
<td>5-40 m/s</td>
</tr>
</tbody>
</table>

![Diagram of nerve fibers and skin layers]
Pathologies

- **large fiber neuropathy**: increased incidence of *carpal tunnel syndrome* (25% of patients ?)
  - sensory neurography
  - motor neurography
  - EMG

- **small fiber neuropathy**
  - sympathetic skin response
  - R-R interval variation
  - laser evoked potentials

*Increase in connective tissue*
Sensory neurography
Sensory neurography
Motor neurography
Motor neurography

\[ \text{vitesse de conduction (m/sec)} = \frac{D \text{(cm)}}{\Delta L \text{(msec)}} \times 10 \]

where $D = \text{distance}$ and $\Delta L = \text{différence de latences}$.
Motor neurography
EMG

A
Effort minime
TRACÉ SIMPLE

B
Effort modéré
TRACÉ INTERMÉDIAIRE

C
Effort maximal
TRACÉ INTERFÉRENTIEL

20 msec

MN
Axone
PLAQUE MOTRICE
JONCTION NEURO-MUSCULAIRE
JNM
m
EMG

Normal
EMG

Myo
Carpal tunnel syndrome
Carpal tunnel syndrome

Ultrasonography
Sympathetic skin response

Efferent part of the SSR reflex: *myelinated sympathetic fibers* from the spinal cord that terminate in paravertebral sympathetic ganglia. Postganglionic fibers are *non myelinated (type C)* and innervate the eccrine sweat glands.

The central part of the reflex arch is not fully understood yet. It is presumably polysynaptic with a connection to the structures of hypothalamus, ventrolateral part of the brainstem, medial and basal parts of the frontal lobe and medial part of the temporal lobe.
SSR can be evoked by different types of stimuli

- electrical stimulation of peripheral nerve in the extremity
- noise
- brisk inspiration
- cough effort
- lie
Sympathetic skin response
R-R interval variation

R-R interval variation is based on the variability of heart rate in a relaxed state and following hyperventilation.

Heart rate variability is the result of the instantaneous relationship between excitatory, i.e., sympathetic, and inhibitory, i.e., parasympathetic, neural influences on the sino-atrial node automatism.
R-R interval variation
# Laser evoked potentials

<table>
<thead>
<tr>
<th>Characteristics of nociceptive stimuli</th>
<th>Electrical</th>
<th>Thermal</th>
<th>Mechanical</th>
<th>Chemical</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>conduct.</td>
<td>radiation</td>
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<tr>
<td></td>
<td>thermode</td>
<td>convent.</td>
<td>laser</td>
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</tr>
<tr>
<td>Specificity</td>
<td>+++</td>
<td>+++</td>
<td>+++</td>
<td>+++</td>
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<tr>
<td>Noninvasive</td>
<td>+++</td>
<td>+++</td>
<td>+++</td>
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<tr>
<td>Reproducibility</td>
<td>+++</td>
<td>±</td>
<td>±</td>
<td>+++</td>
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<tr>
<td>Time locking</td>
<td>+++</td>
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<td>+++</td>
</tr>
</tbody>
</table>

**Laser evoked potentials**

- Electrical: ***
- Thermal: +++
- Mechanical: ---
- Chemical: +++

**Characteristics:**
- Specificity
- Noninvasive
- Reproducibility
- Time locking

**Stimuli Types:**
- Electrical
- Thermal
- Mechanical
- Chemical

**Methods:**
- Conductance
- Radiation
- Thermode
- Conventional
- Laser
Laser evoked potentials

Grand average Cz and T4 (n = 10)
Left hand stimulation

Topographical map
left hand stimulation

Dipolar modeling of late LEPs

N1 160
N2 220
P2 320 (ms)
Laser evoked potentials
Laser evoked potentials

1. Small fiber neuropathies

Electrical stimulation of tibial nerve

- Right: N22, P48
- Left: N22, P48

Laser stimulation of foot

- Right: N22, P48, 2 µV
- Left: N22, P48, 5 µV

Aδ-fiber neuropathy (CRPS)

Controlateral asymptomatic foot

Symptomatic foot

SEP lumbar

SEP scalp

A sural nerve biopsy at the symptomatic foot showed a reduction in Aδ-fiber density of 64%