Pediatric neurophysiology: Neurography and EMG

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Contents:
1. Clinical overview
2. Neurophysiology: Neurography
   EMG
   Other tests
3. Special about investigations in children
DISORDERS OF THE MOTOR UNIT

Motorneuron disease
(SMA)

Neuromuscular transmission disorder
(Cong. myasthenic syndrome)

Polyneuropathy
(Cong. hypomyelination syndrome, GBS)

Myopathy
(Cong. muscular dystrophy, myotubular myopathy, etc.)
PEDIATRIC DISORDERS OF THE MOTORNEURON

- Spinal muscular atrophy (SMA)
- Viral infections, mainly poliomyelitis

**SMA:**
- **Type I** Werdnig-Hoffmann disease; 1/25000 births. Prenatal onset: 30%.
- **Type II** Onset after 3 mos of age.
- **Type III** Kugenberg-Welander syndrome. Insidious onset.
- **Type IV** Adult SMA and Kennedy disease.

Asymmetrical or monomelic SMA. Distal SMA. Bulbar and bulbo-pontine types etc.

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SMA TYPE I

Twin boys 2 m old with feeding difficulties & muscular hypotonus

- No tendon reflexes
- Tongue fasciculations
- Muscle weakness most pronounced in prox. mm

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PEDIATRIC DISORDERS OF THE PERIPHERAL NERVE

- Polyneuropathy
  - Acute - GBS
  - Chronic - HMSN, CIDP, HNPP
- Mononeuropathy
- Plexopathy

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### Some hereditary motor sensory polyneuropathies

<table>
<thead>
<tr>
<th>Type</th>
<th>Usual age of onset</th>
<th>Nerve conduction velocities (m/s)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Demyelinating</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>CMT 1A</td>
<td>1st decade</td>
<td>15 – 20</td>
</tr>
<tr>
<td>CMT 1B</td>
<td>1st decade</td>
<td>&lt; 20</td>
</tr>
<tr>
<td>CMT 1C</td>
<td>2nd decade</td>
<td>16 – 22</td>
</tr>
<tr>
<td>CMT 1D</td>
<td>2nd decade</td>
<td>26 – 42</td>
</tr>
<tr>
<td>CMT 1X</td>
<td>3rd decade</td>
<td>gave eneepications</td>
</tr>
<tr>
<td>HMSN 1</td>
<td>3 years</td>
<td>&gt; 50</td>
</tr>
<tr>
<td><strong>Axonal</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>CMT 2A</td>
<td>10 years</td>
<td>&gt; 30 m/s, reduced amplitudes</td>
</tr>
<tr>
<td>CMT 2B</td>
<td>2nd decade</td>
<td>reduced amplitudes</td>
</tr>
<tr>
<td>CMT 2C</td>
<td>1st decade</td>
<td>&gt; 30 m/s, reduced amplitudes</td>
</tr>
<tr>
<td>CMT 2D</td>
<td>16 – 30 years</td>
<td></td>
</tr>
<tr>
<td>CMT 2E</td>
<td>10 – 30 years</td>
<td></td>
</tr>
<tr>
<td>CMT 2F</td>
<td>2nd decade</td>
<td></td>
</tr>
<tr>
<td>CMT 2G</td>
<td>15 – 25 years</td>
<td></td>
</tr>
<tr>
<td>CMT 2L</td>
<td>15 – 33 years</td>
<td></td>
</tr>
<tr>
<td>HMSN-P</td>
<td>17 – 50 years</td>
<td></td>
</tr>
<tr>
<td>HMSN + ataxia</td>
<td>13 – 27 years</td>
<td></td>
</tr>
</tbody>
</table>

CMT = Charcot-Marie-Tooth, HMSN = hereditary motor sensory neuropathy, HNPP = hereditary neuropathy with liability to pressure palsies, HMSN-P = hereditary motor sensory neuropathy.

**PEDIATRIC DISORDERS OF MUSCLE**

- Congenital muscular dystrophy (CMD)
- Dystrophinopathy (DMD, BMD)
- Sarcoglycanopathy (LGD)
- Structural myopathy (nemaline etc)
- Myositis

**CMD WITH MEROSIN DEFICIENCY**

At some months of age:
- Hypotonia
- Slight facies myopatica

At 2 years of age:
- Sits with support
- Hyperlordosis
- Facies myopatica

CMD - merosin deficiency type

**Patien**

**Healthy control**
Normal merosin

Ex. myopathic motor unit potentials (MUPs)

- Malformations of the brain
- Hydrocephalus
- Microtia and cloudy eyes
- Severe mental retardation
- Myopathy

DMD: Boy 4.5 yrs with Walker Warburg syndrome

DISORDERS OF NEUROMUSCULAR TRANSMISSION

PRESYNAPTIC DEFECTS 9

SYNAPTIC DEFECTS 17

POSTSYNAPTIC DEFECTS 95

Ex. Gene products that mutate in CMS

Neuromuscular Junction

Presynaptic
- ChAT
- Synaptic
- COLQ
- Postsynaptic
- Agrin
- RAPSN
- MuSK
- DOK-7
- SCN4A
- CHRNB
- CHRND
- CHRNA
## Congenital Myasthenic Syndromes

<table>
<thead>
<tr>
<th>Disease</th>
<th>Presynaptic</th>
<th>Postsynaptic</th>
</tr>
</thead>
<tbody>
<tr>
<td>Abnormal ACh release</td>
<td>Respiratory difficulties from birth</td>
<td>Prolonged response to Tensilon</td>
</tr>
<tr>
<td>Abn ACh resynth or mobil.</td>
<td>Ptosis, apnoea, sudden death</td>
<td>Ptosis, ophthalmoplegia, Tensilon variable</td>
</tr>
<tr>
<td>Slow channel syndrome</td>
<td>Limb weakness, Slow progression</td>
<td>Prolonged response to Tensilon</td>
</tr>
<tr>
<td>Impaired function of AChRs</td>
<td>Respiratory &amp; feeding problems, ptosis, strabismus, Tensilon positive</td>
<td>Prolonged response to Tensilon</td>
</tr>
<tr>
<td>Reduced number of AChRs</td>
<td>Respiratory problems from birth, later ptosis &amp; facial weakness</td>
<td>Prolonged response to Tensilon</td>
</tr>
<tr>
<td>Abnormal ACh release</td>
<td>Respiratory problems from birth, later ptosis &amp; facial weakness</td>
<td>Prolonged response to Tensilon</td>
</tr>
<tr>
<td>Endplate AChE deficiency</td>
<td>Ptosis, Negative Tensilon test, Diffuse weakness &amp; ophthalmoplegia</td>
<td></td>
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</tbody>
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**Suspect CMS in infants at:**

- Bulbar palsy
- Vocal cord weakness
- Dysphagia
- Focal contractures
- Floppiness
- Episodic apnoe
- Need of ventilator
- Sib deceased in 'myopathy UNS'

CMS, congenital myasthenic syndrome

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**Dok-7 mutation**

**Probable the 3rd most common form of CMS**

**DOK-7 activates MuSK. Important for the development of endplates. Mutations in DOK-7 give smaller endplates.**

**Proximal muscles more engaged than distal. Normal initial development, thereafter decline. Clinical picture similar to limb-girdle myopathy.**

Does not respond to AchE inhibitor. May respond positively to ephedrin.

Common mutation is 1124_1127 dup THCC at screening.
**Symptoms and signs in some disorders in neonates where weakness and hypotonia predominate**

<table>
<thead>
<tr>
<th>Symptoms</th>
<th>Possible disorder</th>
</tr>
</thead>
<tbody>
<tr>
<td>Respiratory problems</td>
<td>CMD, nemaline myopathy, neonatal myasthenic syndrome, myotonic dystrophy, myotubular myopathy, SMA.</td>
</tr>
<tr>
<td>Difficulty swallowing</td>
<td>As above</td>
</tr>
<tr>
<td>Facial weakness</td>
<td>CMD, myotonic dystrophy, myotubular myopathy</td>
</tr>
<tr>
<td>Fasciculations</td>
<td>SMA</td>
</tr>
<tr>
<td>Contractures</td>
<td>CMD, myotonic dystrophy</td>
</tr>
<tr>
<td>Ptosis/ophthalmoplegia</td>
<td>CMD, neonatal myasthenic syndrome, myotubular myopathy, mitochondrial myopathy</td>
</tr>
<tr>
<td>Dysmorphic features</td>
<td>Nemaline myopathy, myotubular myopathy</td>
</tr>
</tbody>
</table>

**NERVE CONDUCTION STUDIES**

**NCS; Neurography**

- Routinely performed with surface electrodes by biomed. technologists.
- All EMGs are preceded by NCS. In many patients only NCS is performed.
- Motor investigations always include F-wave studies.
- Autonomic and sensory tests are performed when indicated.

**Hand of a neonate**

Motor nerves: 2 stimulation points; distal and proximal.
Sensory nerves: 1 stimulation point.

**Tibial nerve**

- Distal stimulation
- Proximal stimulation
**MOTOR NERVES & PARAMETERS**

Distal motor latency (ms)
Proximal motor latency (ms)
Distal motor amplitude (mV)
Proximal motor amplitude (mV)
Conduction velocity (m/s) = \( \frac{\text{proximal latency} - \text{distal latency} \times 100}{\text{distance (mm)}} \)

F-latency (ms): signal to anterior horn cells and back to muscle.

**NEUROGRAPHY: 1w boy, with chondrodysplasia and hypotonia**

For "screening": nerves of 1 arm and 1 leg examined.

**GBS IN A 2-YEAR OLD GIRL**

Median nerve:
- Conduction block
- Temporal dispersion
- Prolonged distal latency
- Decreased conduction velocity
- Low CMAP
Commonly used muscles:
- Tibialis anterior
- Vastus lateralis
- Biceps brachii
- Deltoid
- Interosseus dorsalis I

Above muscles rather easy to get voluntary activated, and represent proximal and distal parts of upper and lower limbs.

**SPONTANEOUS ACTIVITY**
- Positive sharp waves
- Fibrillations
- Myotonia
- Complex repetitive discharges

**COLLECTION OF MUPs QUANTITATIVELY**

Special reference values on MUP data for children not collected.
COLLECTION OF MUPs SEMIQUANTITATIVELY

- NO AVERAGING
- COLLECTION OF MUPs INTERFERING WITH ONE ANOTHER
- DURATION OF MUPS INEXACT.

INTERFERENCE PATTERN

TA analysis

1 sec

Neuropathy
Myopathy

Collateral sprouting in neurogenic disorders, e.g. SMA, allows for enlarged but fewer MUPs.
Reinnervation potentials - enlarged MUPs due to reinnervation by collateral sprouting

Girl, first child of healthy parents, SMA type I

2-year old boy with DMD

6.5 year old boy with DMD
QEMG: boy 1w old with chondrodysplasia + hypotonia

Normal EMG in right m. tibialis anterior

NEUROMYOTONIC DISCHARGES IN SCHWARTZ-JAMPEL SYNDR. 

Boy, 1 week of age. Parents are cousins.

Severe myopathy in right deltoid muscle

Boy 2 weeks old, with artrogryphosis & amyoplasia & myopathy

Left vastus lateralis
The same boy; artrogryphosis, amyoplasia and myopathy

Dermatomyositis in a 14 year old boy

Myopathic EMG and Neuropathology

The muscle fiber diameters vary in these 3 disorders, with an increase of small muscle fibers. EMG shows in all a myopathic picture of different severity.
12-year old girl with muscular fatigue in extremities and tongue; bilateral slight ptosis

- No extradischarges in motor nerves
- NCS normal
- F-responses: CMAPs stable

Repetitive nerve stimulation:
- ADM
- M. anconeus
- M. nasalis
- M. trapezius

Stimulated SFEMG in the very youngest
REPETITIVE NERVE STIMULATION (RNS)

EDC good muscle for RNS in children. Decrement >10% (3Hz) pathological.

Newborn babies
Neuromuscular synapses immature. Low-frequent stim. does not give decrement or facilitation.
High-frequent stim. (20Hz) may give decrement up to 25%. 5-10Hz stim. may give a slight facilitation.

Children <1 yr & relatively low suspicion of CMS
3Hz stim and thereafter 10 Hz stim, then 5Hz stim again. If normal, SfEMG may be spared.

Abnormal
Increment >20%
Post-tetanic or post-exercise facilitation >120%
Low-frequent stim with decrement >20%

BLINK REFLEXES
Via n trigeminus and n facialis monosynaptic and oligosynaptic tracts through the brain stem are investigated. No special preparation needed.

Stimulation: N Trigeminus supraorbitalis
Recording: M Orbicularis oculi

BLINK REFLEXES
CONGENITAL BILATERAL FACIAL PALSY, Moebius syndrome

Absent blink reflex
Normal blink reflex for comparison
EMLA CREAM FOR SURFACE ANESTHESIA

EMLA = combination of prilocain and lidocain. Applied under occlusion 45-60 m.

DORMICUM® (midazolam) FOR SEDATION

**Administration:**

<table>
<thead>
<tr>
<th>Route</th>
<th>Dosage</th>
<th>Max dosage</th>
<th>Effect after</th>
<th>Duration</th>
</tr>
</thead>
<tbody>
<tr>
<td>RECTAL</td>
<td>0.2-0.3 mg/kg</td>
<td>5 mg</td>
<td>10-20'</td>
<td>30-60'</td>
</tr>
<tr>
<td>NASAL</td>
<td>0.2 mg/kg</td>
<td>5 mg</td>
<td>4-5'</td>
<td>20-30'</td>
</tr>
<tr>
<td>ORAL</td>
<td>0.3-0.5 mg/kg</td>
<td>7.5 mg</td>
<td>15-30'</td>
<td>30-50'</td>
</tr>
<tr>
<td>IV</td>
<td>0.1 mg/kg</td>
<td>5 mg</td>
<td>2-3'</td>
<td>15-30'</td>
</tr>
</tbody>
</table>

**Relative cons:**

MG, SEVERE NEUROMUSCULAR DISORDER WITH RESPIRATORY FAILURE, HYPOXIA, RESPIRATORY FAILURE UNS, RENAL FAILURE.

**Conclusions**

- Although we live in the era of DNA analyses, neurophysiological methods in the work-up of neuromuscular disorders are of basic importance.
- Disorders in different parts of the motor unit are investigated.
- Electrodes and preparations are adjusted to the pediatric patients, who need to be examined in the presence of their parent/s. Be quick and slick as well as angelic in patience.